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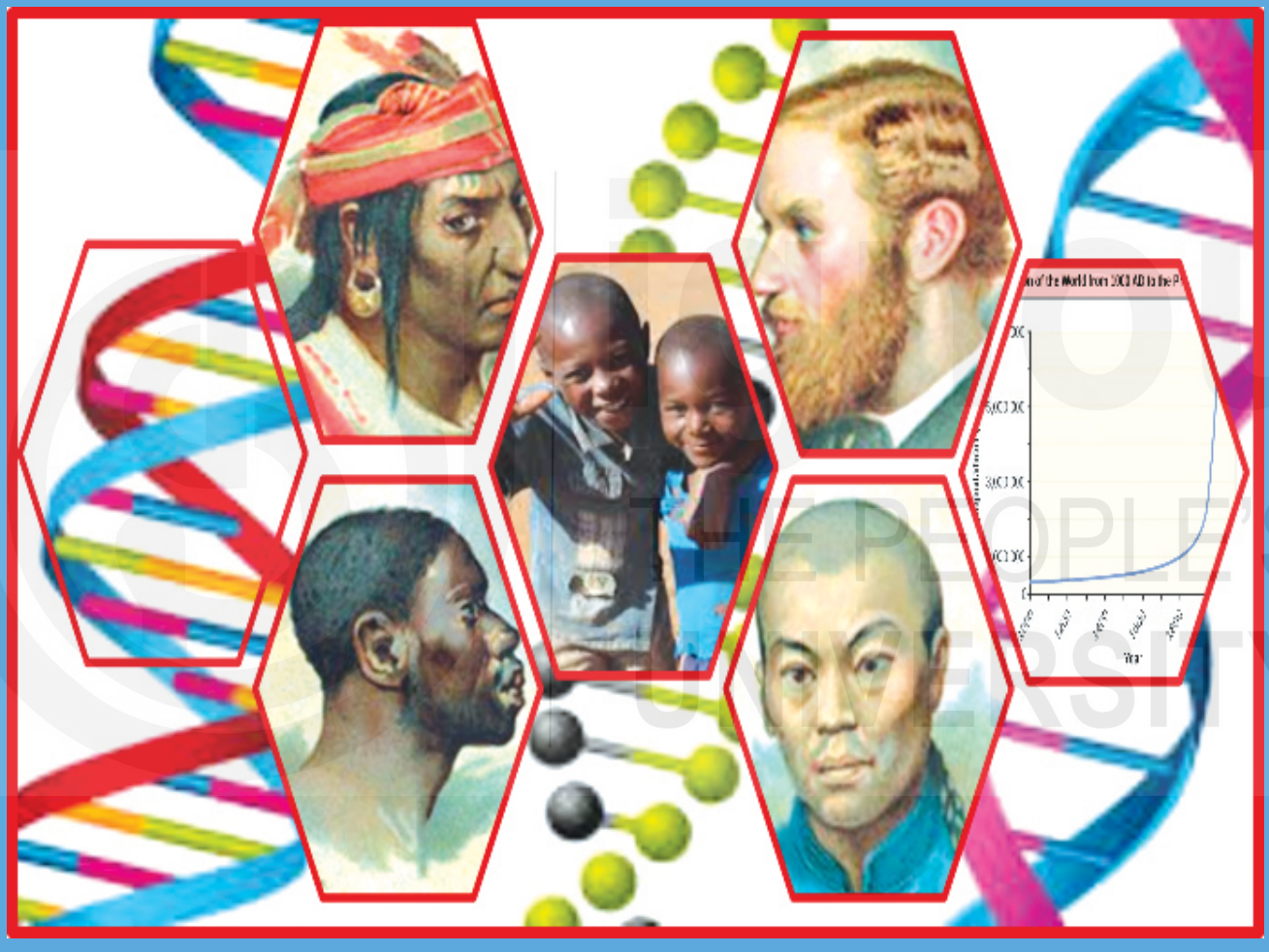
School of Social Sciences

BANC-107
Biological Diversity in Human Populations



"शिक्षा मानव को बन्धनों से मुक्त करती है और आज के युग में तो यह लोकतंत्र की भावना का आधार भी है। जन्म तथा अन्य कारणों से उत्पन्न जाति एवं वर्गगत विषमताओं को दूर करते हुए मनुष्य को इन सबसे ऊपर उठाती है।"

– इन्दिरा गांधी

"Education is a liberating force, and in our age it is also a democratising force, cutting across the barriers of caste and class, smoothing out inequalities imposed by birth and other circumstances."

– Indira Gandhi

BANC-107

BIOLOGICAL DIVERSITY IN HUMAN POPULATIONS

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COURSE INTRODUCTION

Physical or Biological Anthropology is one of the four main branches of Anthropology encompasses the pattern of variation at population level by understanding the physical and genetic traits. Thus, the course mainly focuses to understand the population in total from micro to macro level including genetic traits to races. This will allow the readers to equip themselves at the end of the course to find out the variations that precipitate in terms of abnormalities.

Course Presentation

The course, Biological Diversity in Human Populations consists of three blocks of theory and one practical manual.

Block-1 (Unit 1 through Unit 4) provides a general introduction to biological anthropology in understanding variation from culture to genetics. Unit 1 introduces the significance of biological anthropology in studying biological diversity using phenotypic and genetic markers. Unit 2 describes the basis of genetic variation. Unit 3 discusses the distribution of various genetic markers of blood (Serological, Biochemical and Molecular) in Indian populations. Unit 4 focuses on the role of the influence of Bio-cultural factors on diseases and diet.

Block-2 (Unit 5 through 7) explains the major races of the world. It also provides a detailed description on the classification of the people of the world and India. Unit 5 describes the classification of Indian populations by various scholars like Risley, Guha, Sarkar and Balakrishnan. Unit 6 focuses on the racial elements that existed during pre and proto historic periods. Unit 7 provides the detailed description on major races (Caucasoid, Negroid and Mongoloid) of the world.

Block-3 (Unit 8 through 10) covers in detail about the demography particularly emphasising Indian scenario and inbreeding effects on demography. Unit 8 describes the concepts of Demography. In Unit 9, the focus will on Indian Demography. Unit 10 discusses the consequences of inbreeding in Indian populations.

The Practical Manual describes craniometry, serology and dermatoglyphics. The practical manual also insists demographic data. Craniometry covers various measurements on Skull and Mandible. Serology describing the procedures of determination of A1, A2, B, O; MN; and Rh (D) blood group systems. Then Finger and Palmar Dermatoglyphics shall also be covered. Finally, procedures for collecting the Demographic data through secondary sources is presented.

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Block 1

INTRODUCTION TO BIOLOGICAL DIVERSITY

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UNIT 1 IMPORTANCE AND IMPLICATIONS OF BIOLOGICAL VARIATION*

Contents

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1.1 Physical Anthropology vs. Biological Anthropology

1.2 Categorisation of People

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- 1.2.2 Race
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- 1.3.1 Somatoscopic Characters
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1.5 References

1.6 Answers to Check Your Progress

Learning Objectives

After reading this Unit, you would be able to:

- Understand the definition and branches of anthropology;
- Explain how the people are categorized into races and ethnic groups; and
- Appreciate the use of morphological, and genetic characters in understanding biological diversity.

*Contributed by Dr. P. Venkatramana, School of Social Sciences, IGNOU, New Delhi.

1.0 INTRODUCTION

Anthropology has been called the holistic study of humankind. In simple words the subject matter of Anthropology deals with human evolution and variation (diversity). Here humans not as individuals but group of people or populations are a unit of study. As stated, investigation of human diversity in time and space has been one of the core aspects of Anthropology. Anthropology examines the human diversity in respect of biology, society, language, and culture.

The anthropology includes four main branches: Social-cultural anthropology, Physical/Biological anthropology, Archaeological anthropology and Linguistic anthropology.

A brief discussion of the four branches is as follows:

Social-cultural anthropology: This branch deals with the patterns of belief and behaviour in past and modern cultures. The origin of this branch can be traced back to the nineteenth century, when travel and explorations brought into contact with diverse cultures of Africa, Asia, Oceania, and the New World. It deals with human society (social and political organizations) and cultures (human behaviour, knowledge, thoughts, beliefs, customs, art, and law etc). This branch of anthropology is called as social anthropology in England and termed as cultural anthropology in United States of America and in 19th century known as ethnology. Socio-cultural anthropology is a comparative discipline attempts to investigate the similarities and differences across human societies. As a systematic science began when Europeans developed colonies and inducted their culture. Earlier studies conducted by diplomats, travellers and missionaries. The theoretical framework of this subject was developed during 17th century and contributed by Thomas Hobbes, Levitan and Herbert Cherbury. Evolutionary theory in socio-cultural anthropology was proposed by Edward Tylor during 19th century known as unilinear cultural evolution. Using this theory as framework an attempt was made to understand the cultural history of mankind. After unilinear cultural evolution, Franz Boas propounded historical particularism, Bronislaw Malinowski initiated functionalism, Radcliff Brown explained the concept of structural functionalism, Ruth Benedict, Margaret Mead, Linton, Cora-Du-Bois brought thought as known as culture and personality school, V.Gordon Child, Julian Steward and Lesli White introduced cultural ecology and neo-evolution, Claude Levi-Strauss developed the concept of structuralism and Emile Durkheim, Clifford Geertz, Mary Douglass and Victor W Turner proposed symbolic and interpretive anthropology. Presently socio-cultural anthropologists are involved in the studies of human diversity in time and space and addressing contemporary issues and the areas of research such as globalization, diaspora, multiculturalism and transnationalism are emerging area in this sub discipline of anthropology.

Physical anthropology: The primary objective of physical anthropology is to study human biological evolution and variation. Physical anthropology studies biology and behaviour of human beings, non-human primates, extinct hominids, their variation and importance. In view of more emphasis on biological problems by physical anthropologists this sub discipline of anthropology is also termed as biological anthropology. The main focus

of physical anthropology is human evolution, human variation and human adaption in different geographical settings. Physical anthropology developed as a systemic science in 19th Century. Blumenbach who livid during the period of 1752-1840 is credited as Father of Physical Anthropology and who made major contribution in Craniology. When we turn the page of history of physical anthropology, we notice that initially physical anthropologists laid interest on investigating human variations by classifying into different races, physical variation using anthropometry and variations in skin colour, form and colour of hair and eye using somatoscopic observations, primate anatomy and behaviour. Developments in molecular biology such as polymerase chain reaction and gene/genome sequencing led the physical anthropologist to employ molecular tools such as single nucleotide/restriction fragment length polymorphisms, microsatellite markers and haplotypes to draw inferences on origin of human species, gender specific migrations and evolutionary relationships among human populations. Palaeoanthropology (focus on human evolutionary studies), paleo-primatology (study fossil primates), osteology (devoted to the study of bones), human genetics (deals with human hereditary and variation), population genetics (investigates process of evolution), molecular anthropology (study relationship between present day human populations with extinct human remains using DNA), human biology (involved in the study of human variation), human growth and development (emphasize on human growth and development), human ecology (explore relationship between humans and their environment), forensic anthropology (identify skeletal remains for legal cases) and demography (study fertility and mortality, age-sex structure, migration and spatial distribution) are the important areas of study for the physical anthropologists.

Archaeological anthropology: This branch dealswith the origins and development of humankind prior to the invention of script. Archaeological anthropologists scientifically recover, analyze and interpret the material remains of past societies. The subject matter of Archaeological anthropology include prehistory, protohistory and civilizations of the remote past.The archaeological anthropologists arrive at conclusion by examining human, animal and plant remains and material products such as hearts, pottery, tools and associated enclosures. The main methods employed by archaeological anthropologists included excavation, survey and data analysis of available evidence. Archaeological anthropologist attempts to reconstruct the life style of the past people without evidence or undeciphered material and report the origin, growth and development of past cultures. Earlier archaeological anthropologists were more focussed on dating the physical and cultural remains of the past now shifted their attention to economy, subsistence pattern and environment. The term prehistoric was proposed by Daniel Wilson in 1851. Prehistory is period divided into stone age, copper/bronze age and Iron age. Stone age was further classified into Palaeolithic (lower, middle and upper), Mesolithic and Neolithic. Protohistory falls between prehistory and history and dated during the period of 3500-3000 BCE. In India the Indus Valley Civilization falls under protohistoric period.

Linguistic anthropology: This branch devoted to the study of human languages. It focuses on the study of human speech and language and examines the origins of languages in general and specific languages in particular. Linguistic anthropologists examine the association between present-day languages

and trace historical relationship between particular languages and group of languages. Language is an integral part of human behaviour and vehicle of cultural transmission. The scope of linguistic anthropology encompasses exploring the relationship of language and culture, emergence and divergence of languages passing time, origin, evolution and development of languages, languages on the verge of extinction, effect of language on social life, past, present languages and unwritten languages. Franz Boas was the first to emphasize the linguistic aspects of culture. Edward Sapir (1884-1939) coined the term 'anthropological linguistics'. In 1960 linguistic anthropology came into existence in United States. Both linguistics and linguistic anthropologists study language. Linguistic anthropology deals with the relationship of language and cultural behaviour. Linguistic anthropology is divided into historical (study emergence a divergence of languages) and structural linguistics (investigate the role of language in social behaviour). It was observed that the pattern of speech differs across depending on the action, behaviour and communication. Language socialisation (rearing and teaching of individual the basics of social life and its various aspects) and language ideology (exploring the relationship of language and social, economic and political ethics of society) are present subjects of linguistic anthropology.

1.1 PHYSICAL ANTHROPOLOGY VS BIOLOGICAL ANTHROPOLOGY

Physical anthropologists in the beginning have used the anthropometric and anthroposcopic data to describe and classify humans into various population groups and were focused on four limited areas of research such as body dimension, human evolution, race and cranial dimensions.

Physical anthropology is also called as biological anthropology. The biological anthropology reflects the change in the importance to more biologically oriented topics, such as genetics, evolutionary biology, nutrition, physiological adaptation, and growth and development. This shift has occurred since the late 1950s largely because of advances in the field of genetics and molecular biology.

The following areas are the areas of interests of biological anthropologists in understanding the biological diversity:

Human genetics, human evolution, human growth and development, body composition, human biological adaptation to stresses like heat, cold, and altitude and social life of monkeys, apes, and other non-human primates.

1.2 CATEGORISATION OF PEOPLE

Human beings exhibit significant individual variation, but anthropology deals with variation within and between populations.The subject matter of anthropology deals both biological and cultural variations observed between different human populations. Human biological diversity is extensive field and can be studied with the help of morphological, anthropometric and genetical characters.

In modern times, human groups are studied in terms of populations, ethnic groups or races. Therefore, the definitions for the Race, Ethnic Group and

Population are given below:

1.2.1 Population and Mendelian Population

A population can be defined as a group of interbreeding individuals. More specifically, a population is the group within which a person is most likely to find a mate in the cultural group of his/her birth. As such, a population is characterized by a degree of genetically relatedness and shares a common gene pool. Hence a tendency for greater genetic resemblances among persons within populations observed than persons from different populations of the species.

Human evolution is a significant component of biological anthropology. From the evolutionary point of view, the unit of study is a population. In genetics jargon, population, i.e., Mendelian population is a technical term. Consequently, a Mendelian population is a group of people who reproduce sexually or are potentially capable of doing so and sharing a common gene pool, focuses on the evolutionary aspect. An evolutionary biologist Theodosius Dobzhansky shas defined mendelian population as "a reproductive community of sexual and cross fertilizing individuals which share in a common gene pool"

Theoretically, the concept of the Mendelian population may be extended to include the entire human species, because all humans share the common gene pool of the species Homo sapiens. In this sense, mankind itself constitutes a "population" and can be considered a proper unit of genetic study. The term deme is often used instead of the term population, with reference to a small endogamous group which is relatively self-sufficient and isolated from other such groups. Demes are the smallest basic population units studied by the population geneticists, but they can be organized into complexes of increasing size and inclusiveness-tribes, states, national and supranational entities-until all the demes of mankind are included.

Check Your Progress

1) Distinguish between population and Mendelian population.

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……...……

1.2.2 Race

Various scholars have defined the term 'race' in 20th century in various ways which gave more or less similar meaning. One of the definition of race given by E.A. Hooton (1926) is as follows: "A race is a great division of mankind, the members of which, though individually varying, are characterised as a group by a combination of certain morphological and metrical features, principally non-adaptive, which have been derived from their common descent".

However, Ashley Montague (1905-1999), an anthropologist opposed the use of the term race and proposed the term ethnic group to describe human populations, though he did not invent the term ethnic group. However, in a contemporary biological sense, races can be seen as subspecies, a group of populations within a species that are thought to have a much closer shared ancestry within the race than with populations that are grouped in other races.

Fig. 1.1: The racial diversity of Asia's peoples
(https://en.wikipedia.org/wiki/Race_(human_categorization)#/media/File:Asiatiska_folk,_Nordisk_familjebok.jpg)

1.2.3 Ethnic Group

An Ethnic group or Ethnicity is a category of people who identify with each other based on common language, ancestral, social, cultural or national experience. An ethnic group may include a collective name, belief in common descent, a sense of solidarity, and an association with a specific territory, which the group may or may not hold (*Ryan,* 1990).

In common practice, race is a vague term that so often is used interchangeably with ethnic group. Sometimes, few consider the two terms, race and ethnic group synonymous even though they are not. However, race and ethnicity are strongly related in certain ways. Both race and ethnicity are used for categorizing people. Anthropologists use physical characteristics to define a "race" and cultural or historical and geographical characteristics for ethnicity.

Fig.1.2: Multi ethnic group of people, census
(Source: https://www.ppic.org/topics/trending-2020-census/multi-ethnic-group-of-people)

A detailed discussion on the concept of race and ethnicity are presented in units 5, 6 and 7 of this course.

1.3 HUMAN BIOLOGICAL VARIATION

A significant and the basic concept of physical/biological anthropology has been the study of human biological variation. Human beings across the globe differ from each other in respect of many biological characters. To understand biological variation, somatometric and somatoscopic variables were used by anthropologists from 19th century. These characters were also used throughout the world to describe humans and to classify them into different races. Further the Dermatoglyphics were also used in describing ethnic and/or population variation, it was rarely used in the racial classification. Then beginning of twentieth century marked by the discovery of genetically determined ABO blood group system followed by other blood group polymorphisms, protein polymorphisms, red cell enzyme polymorphisms and recently DNA markers offer anthropologists with tools to study human variation.

Therefore, a brief account of morphological (*somatoscopic, somatometric, dermatoglyphics*) and genotypic (serological and molecular) characteristics are discussed below with examples:

1.3.1 Somatoscopic Characters

Somatoscopy is the description of morphological characters of humankind. Somatoscopy is the systematic visual observation of physical features of different parts of human body for accurate description. These morphological characters are quantitative in nature hence descriptive in approach. Most somatoscopic characters show geographical variation. Hence these morphological characters were used by Anthropologists to classify human populations. Characters such as skin colour, hair colour, hair form, hair texture, hair whorl, nose form, face form, which are considered in this unit, are described as under:

1.3.1.1 Skin Colour

Broadly three shades of skin colour are found in human beings. They are: white skin, yellow skin and black skin. Most white skinned people are found in Europe. Mongoloids have yellow skin and people from African countries have generally black skin.

1.3.1.2 Hair

This somatoscopic character includes hair colour, hair from, hair texture and hair whorl.

Hair colour: There is considerably less variability in human hair colour than in skin colour. Different hair colours are seen across the globe. Fisher-Saller have prepared a colour chart with hair samples of thirty different shades. All the thirty shades fall in to three broad categories: Blond, Dark Brown and Red. The range of the hair colour among the Indian population is categorized as light brown, medium brown, dark brown and black.

Hair form: Form of hair may broadly be divided into three types viz., Straight hair (*Leiotrichy*), wavy hair (*Cymotrichy*) and Woolly hair (*Ulotrichy*).

Leiotrichous hair (straight hairs) include subtypes such as *stretched* (usually straight), *smooth* (thinner and flatter), and *flat wavy* (hair has the tendency to become wavy). Similarly, wavy hair also includes three subtypes such as, *broad wave, narrow wave and curly*. Of these three, both broad and narrow wavy types lie on the same plane, but the curly type dose not because of large spirals. Woolly hair (*Ulotrichy*) may be of different types such as *Frizzly*-waves with very strong curvature; *Loose frizzles* – circular and flat spirals (about 1.5 cm dia.); *Thick Frizzles*- flat spiral hair (less than mm dia.); and *Filfil*– small knots of thick rolled hair (Peppercorn). Various hair forms are depicted in Figure 1.3.

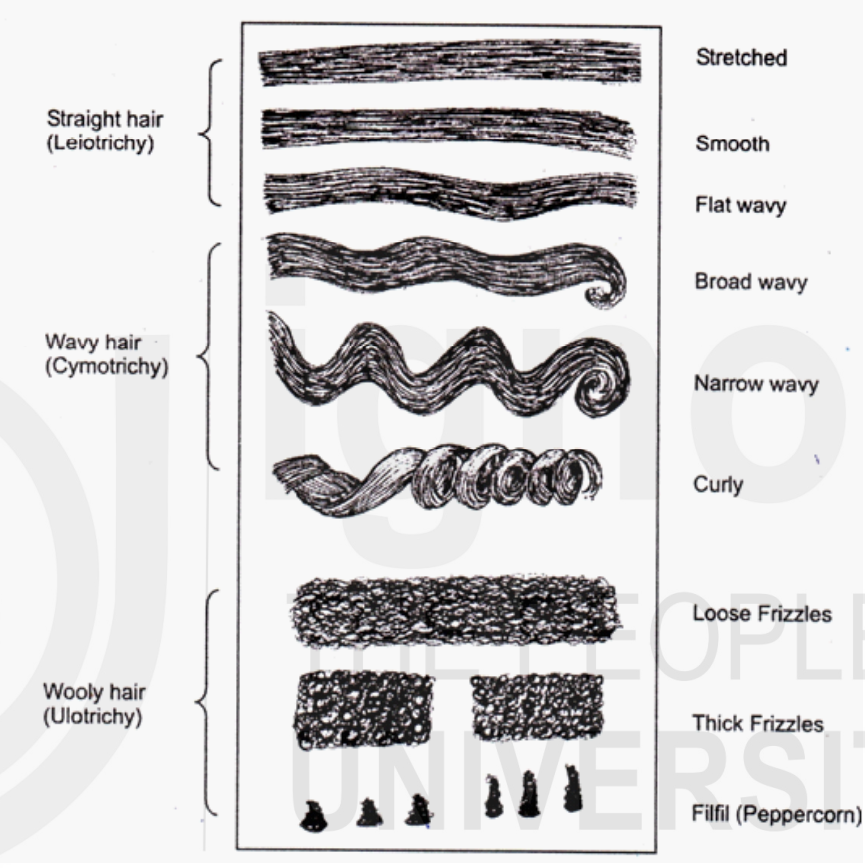

Fig.1.3: Different types of hair form

Hair texture: Hair texture are classified into fine, medium and coarse.

Hair whorl: Hair whorls are usually found on the occipital region (back of the head). Whorls are very rarely found in front portion of the head. Whorls are of two types, clockwise and anti-clockwise.

1.3.1.3 Eyes

Somatoscopic observation on eyes include colour of the iris, eye fold and direction of the eye. Colour of the iris may be classified as black brown, dark brown, brown, light brown, greenish, grey, light grey, dark blue and light blue.

Eye fold: In this somatoscopic character, presence or absence of eye fold is considered. A common variety of eye fold is 'epicanthic fold'. In epicanthic fold, the fold covers the free edge of the inner angle of the eye and may extend on to the cheek (Figure 1.4).

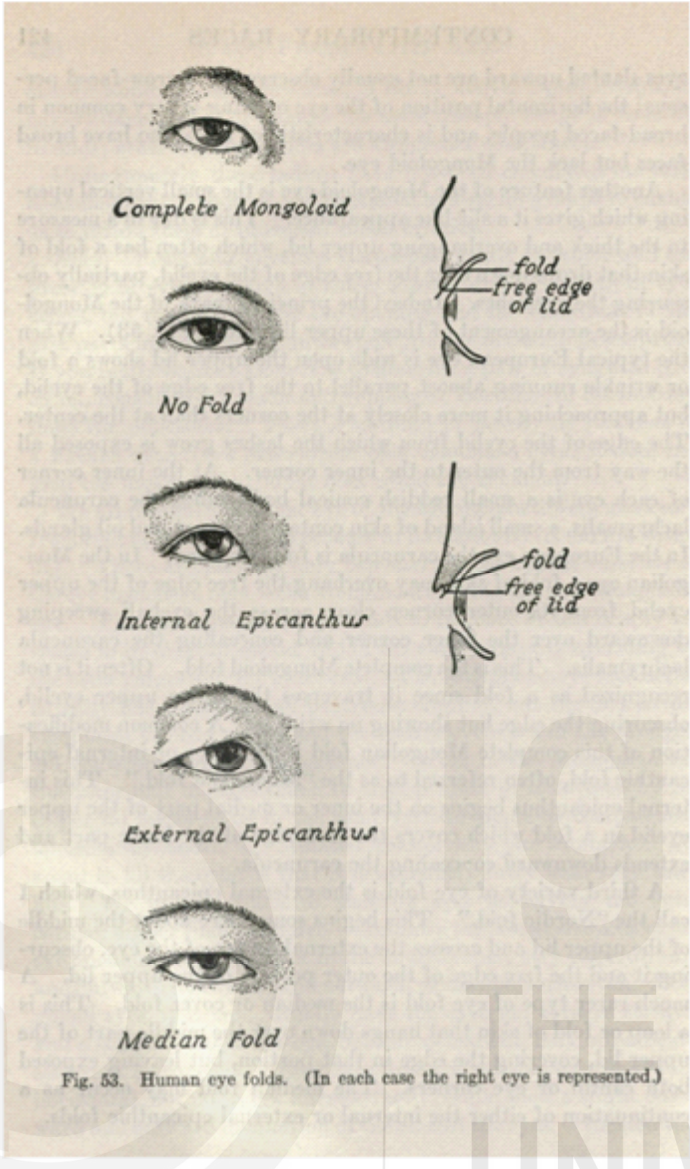

Fig. 53. Human eye folds. (In each case the right eye is represented.)

Fig. 1.4: Eye fold pattern
(Source: s1.zetaboards.com).

Check Your Progress

2) What is Epicanthic fold?

……

……

1.3.1.4 Nose

Different features of nose such as depression of root, nasal profile, nasal septum, nasal bridge, nasal tip, nasal wings are worth studying. The tip of the nose can be upwards or downwards and the profile could be rounded at point or fully rounded or flat. The root of the nose may be recorded as narrow, medium or broad; from the side view many appear depressed which again may be shallow, medium or deep or absent. The nasal bridge may be recorded as straight, concave (slight, medium, markedly), convex- (slight, medium, markedly) or wavy (slight, medium, markedly). The size of the nasal bridge may be narrow, medium or broad. Various types of noses in profile are shown in Figure 1.5.

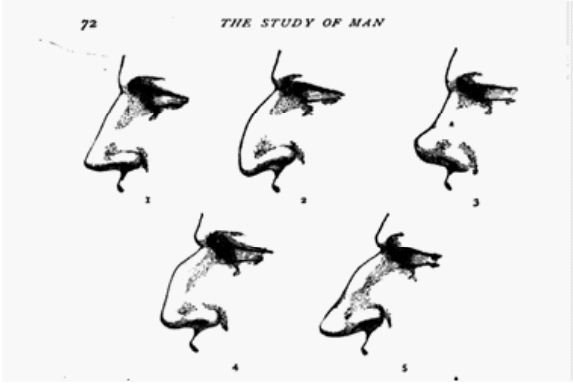

Fig. 1.5: Nose form
(Source: s1.zetaboards.com).

1.3.1.5 Lips

The thickness of the membranous lip is studied with the best observation in profile view. It may be thin, medium, thick and puffy with convex profile, above which the integument lips are deeply concave. The degree of eversion of membranous lip may be absent, moderate, or marked (as in people of African Ancestry).

1.3.1.6 Face

The face can be described in terms of height (long, medium or short), diameter (narrow, medium, broad and very broad), shape, malar prominence and prognathism. The shape may be oval, elliptical, round, square, quadrangular or flat (Figure 1.6). Prominence of the cheek bone (malar) is an important feature; it is described as absent, slight, moderate or marked. Alveolar protrusion of face is called prognathism. Profile view is best to ascertain it to be slight, moderate or marked.

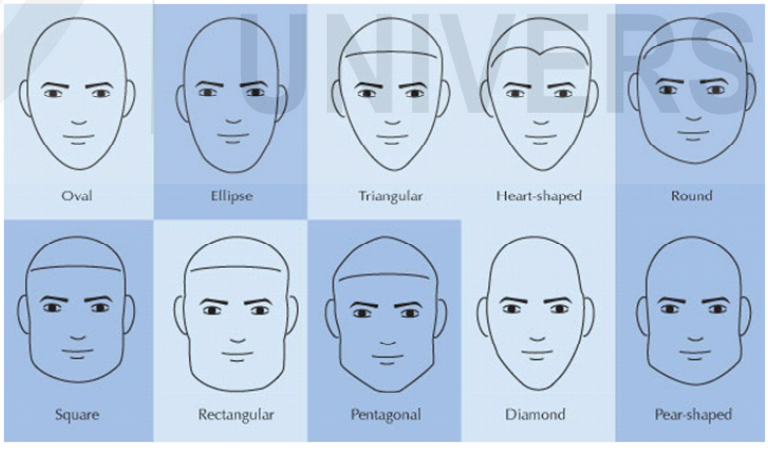

Fig. 1.6: Different shapes of face
(Source: https://design.tutsplus.com/tutorials/human-anatomy-fundamentals-advanced-facial-features--cms-20683)

1.3.2 Anthropometric Characters

Anthropometry is a major technique of physical anthropology. Anthropometry is a method to take measurements of human body. It is the means of quantifying variation in body size and shape. It may be defined as the systematic recording of measurements on human being both living and dead. Anthropometry can be

divided into Somatometry and Osteometry. Somatometry is the recording of measurements on living body or cadaver, including head and face. Osteometry is the measurements on skeleton, including Craniometry, which deals with measurements on skull. The somatometric measurements includes; height, sitting height, body weight, head circumference, chest circumference, abdominal circumference, head length, head breadth, skinfold thickness measurements such as triceps, biceps, subscapular, etc.Osteometric measurements includes maximum length of radius, maximum length of ulna etc. Craniometric measurements includes, maximum cranial length, maximum cranial breadth, nasal height, nasal breadth etc.

But for the present unit, we shall study only somatometry. These somatometric measurements have been used in understanding population variation. The measurements like height vertex (stature), two indices (cephalic index (CI) and nasal index) and their distribution in Indian populations are discussed as under:

1.3.2.1 Stature

Stature (standing height) measures the vertical distance from the standing floor to the vertex. Here vertex is the highest point on the head when the head is in the Frankfurt-Horizontal (FH) plane. Vertex is not an anatomically fixed point and is dependent on the orientation of the head.

Stature can broadly be classified into three categories; tall, medium and short. The height of the tall people ranges from 168-172cm; medium statured people ranges from 158-168cm and short statured people ranges from 148-158cm.

Distribution: The available data on the stature (standing height) among the Indian populations explains that in general caste populations are taller as compared to scheduled castes while tribal groups are short in stature. When the height is examined locality wise, people from Punjab, Delhi, and Rajasthan are in general taller than the populations of other regions, which show a gradient of stature distribution from north-west to east and south positions of Indian sub-continent. The populations who speak Austro-Asiatic, Tibeto-Chinese, and Dravidian languages are shorter in stature as compared to the population who speak Indo-European language. On the whole the mean stature of Indian population is medium with 163.03 cm (*Bhasin,* 2006 &2009). The stature among the tribal populations of India ranges from 150.00-162.00 cm.

1.3.2.2 Cephalic Index

By measuring maximum head length and maximum head breadth, we can estimate Cephalic Index (CI) as:

$$\frac{Maximum\ head\ breadth}{Maximum\ head\ length} X100$$

Based on cephalic index people can be classified into:

Hyperdolicocephalic (very long and narrow)	up to 69.9
Dolicocephalic (long and narrow)	70.0-75.9
Mesocephalic (medium)	76.0-80.9

Brachycephalic (short and broad)	81.0-85.5
Hyperbrachycephalic (very short and broad)	85.6and over

Distribution: In Indian populations the mean value of cephalic index is 76.06, which is mesocephalic. The cephalic index ranges from dolichocephalic to mesocephalic except a few populations from West, East, and South India. However brachycephalic has been observed among the populations who live in Islands. When we see the distribution of cephalic index according to zones, dolichocephalic is found in north and central India and brachycephalic is absent in both these zones. However, mesocephalic is found among the west, east, and south Indian populations to a larger extent. It was observed that mean value of cephalic index is low in scheduled tribe populations compared to other ethnic groups. Tribal populations from northeastern and southern region were dolichocephalic (70.0- 75.9), whereas the tribal populations of western region were mesocephalic(76.0-80.9).Also, the scheduled tribe populations with Mongoloid features from Eastern Himalayan region were mesocephalic (Bhasin, 2006 &2009). The mean value of cephalic index in tribal populations is (77.31) (Mesocephalic).

1.3.2.3 Nasal Index

Another important index that is used in population variation is nasal index. It can be obtained by measuring nasal length and nasal breadth and can be calculated as below:

Nasal Index: $\frac{Nasal\ Breadth}{Nasal\ Length} X100$

By using this index, population can be categorised into the following five types:

Hyperleptorrhine (very narrow nose)	up to 54.3
Leptorrhine (Narrow nose)	55.0 — 69.9
Mesorrhine (Medium nose)	70.0 — 84.9
Platyrrhine (broad nose)	85.0 — 99.9
Hyperplatyrrhine (Very broad nose)	100.0 and over

Distribution: Mesorrhine (70.0-84.9) is most predominant among the Indian populations followed by Platyrrhine (85.0-99.9). Among scheduled tribes, the value of the index is high (99.9) as compared to other ethnic groups. The nasal index among the scheduled tribes from Eastern Himalayan region with Mongoloid characters is low (76.69). The value is high among the speakers of Austro-Asiatic and Dravidian languages as compared to Indo-European and Tibeto-Chinese language speakers (*Bhasin,* 2006 & 2009).

Check Your Progress

3) Whataretheanthropometriccharactersthatareusedinstudyingpopulation variation?

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……...……

1.3.3 Genetic Markers

Besides the traditional morphological traits, genetical traits such as ABO, MNSs, and Rh blood groups also exhibit population variation in terms of allele frequencies. All these blood group systems come under genetic markers. We shall discuss genetic markers as under:

1.3.3.1 Serological Markers

The Blood group systems are a classical example for genetic markers. The blood group systems have been studied by Anthropologists to understand population variation and in the classification of human races. A number of blood group systems were discovered. The following table depicts the list of Blood group systems.

Table 1.1: Major Blood Group Systems

System	Year of Discovery	Discoverer
ABO	1900	Landsteiner
AB	1902	Decastello and Sturli
Rh (D)	1940	Landsteiner & Weiner
MNSs	1927	Landsteiner & Levine

Besides the above blood group systems, there are other blood group systems like P, ABH, Lutheran, Deigo, Duffy, Kidd and Kell. But ABO and Rh (D) systems were extensively investigated by Anthropologists and population biologists to understand the population variation within and between populations. The ABO and Rh (D) blood groups and its products are routinely being used in transfusion medicine.

1.3.3.1.1 ABO system

Blood groups A,B, and O were discovered by Karl Landsteiner in 1900 while the AB blood was discovered by Decastello and Sturli in 1902. ABO (ABO, alpha 1-3-N-acetylgalactosaminyltransferase and alpha 1-3-galactosyltransferase) gene variation is responsible for the ABO blood group.ABO gene locus has three alleles: A, B and O, each allele determines the respective blood group. O blood group is due to the mutation of amino acid guanine at the amino terminus of protein, whereas expression of glycotransferase activity of A,B or AB alleles convert the H antigen into A, B or AB antigen.

Distribution: Generally, in Indian populations the frequency of allele O is more than 50% but the frequency of allele B is higher (0.233) as compared to allele A (0.186). Among different ethnic groups the allele B is higher (0.230-0.248) as compared to allele A (0.173-0.181). Among scheduled tribes where alleles A and B are almost similarin frequency(0.213 and 0.218, respectively) (*Bhasin,* 2009).

Check Your Progress

4) What is the general allele frequency distribution of ABO system in Indian populations?

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……

1.3.3.1.2 RH (D) system

The RH (D) blood group system was discovered in 1940 by Landsteiner and Weiner. RHCE and RHD genes encode Rh antigens. Phenotypes of this system are RH D+ and RHD-, Cde, cde,cDe, cDE, CDE, Cde, CdE and cdE. RHD+ is encoded by RHD gene. A mutation in RHD gene results in RHD-phenotype. Other phenotypes are encoded by RHCE gene. Fisher and Race,1962, Weiner and Wexler, 1963 and International society blood transfusion proposed classification of antigens. Fisher and Sanger nomenclature/classification is known as CDE and assumes that antigens (C,D,E,c,d and e) are encoded by gene D,C,E which are inherited together. As a result,eight gene complexes or haplotypes (Dce, DCe, DcE, DCE, dec, dCe, dcE and dCE) are possible which can form 36 possible genotypes.

Distribution: Among the Indian populations the frequency of allele D varies from 0.532 to 1.000. However, the populations with Mongoloid affinities from Eastern Himalayan region found to be with highest frequency (0.938). In different ethnic groups allele 'D' is in high frequency among scheduled tribes from western, north-eastern and southern peninsular regions as compared to other ethnic groups (Bhasin, 2009). When we see the frequency distribution within different tribal population groups, the frequency of "D" gene is found to be 100% followed "d" gene (0-30%).The range of frequencies of various haplotypes of Rh blood group system in Indian populations are as follows: cde(0.00-0.44), cDe(0.00-0.41); CDE(0.00-0.13); Cde(0.00-0.17);CdE(0.00-0.03) and cdE(0.00-0.09) (*Bhasin,* 2009).

1.3.3.1.3 MNSs System

The MNSs blood group system was first discovered by Landsteiner and Levine in 1927.There are more than 40 antigens in the MNSs blood group system. However, the frequently found antigens in the human population are M, N, S, s, and U. M and N antigens (Landsteiner and Levine, 1927) are encoded by Glycophorin A gene while S, s and U (identified 1947 and 1951, respectively) are encoded by Glycophorin B gene (Reid, 2009). The system consists of two co-dominant alleles designated M and N. There are Haplotypes (combination of two or more alleles) in this system; they are MS, Ms, NS and Ns. The common phenotypes in this system are M+N+S-s+, M+N+S+s+, M-N+S-s+M+N-S+s+M+N-S-s+, M-N+S+s+, M+N- S+s. The haplotypes in MNS system in Indian populations reported were MS, Ms,NS and Ns.

Distribution of MN Alleles in Human Populations

Many human populations have approximately equal numbers of M and N alleles, and thus allele frequencies for both are near 0.5. However, in many East Asian and Native American populations the frequency of the M alleles is high, ever above 0.9 in some cases, while among Melanesians and aboriginal Australians, frequencies of M alleles are low, usually below 0.2 (Birdsell 1972). In Indian populations, the range of frequency of various alleles and haplotypes are as follows: M(0.64); N(0.35); MS(0.19); Ms(0.43); NS(0.09) and Ns(0.26) (Mourant et al.1976; Bhasin, 2009).

Another genetic marker that has been used in population variation studies is Dermatoglyphics.

1.3.3.2 Dermatoglyphics

Dermatoglyphics is the study of the epidermal ridge patterns of the skin of the fingers, palms, toes and soles. Every individual possesses distinct features of ridges and their pattern in fingers, palms and soles. The ridge patterns are stable throughout life and are not modified by environmental factors. The patterns are unique to each individual. Because of these qualities these play a very important role in the personal identification, crime detection, twin diagnosis, racial variation and also found to be associated with various diseases and syndromes.

The ridge configuration present on the palm is called Palmar Dermatoglyphics and the ridge configuration present on the fingers is called Finger Dermatoglyphics. The epidermal ridges form definite local design on the terminal segment (phalanges) of digits and also on the palm and toes.

The present unit is devoted to the role of dermatoglyphics in population variation. Hence variation in Finger Dermatoglyphics is presented.

Finger Dermatoglyphics refers to the configurations present on the distal phalanges of fingers and toes. Based on the construction, Galton (1891), classified them as Arches, Loops and Whorls. Later on, Henry (1900) proposed a four-fold classification. He classified the type as arch, loop, whorl and composite.

Finger Dermatoglyphics are of different types. They are:

Arch

Plain Arch

Tented Arch

Loop

Radial Loop

Ulnar Loop

Whorl

Plain/True Whorl

Composite

Central Pocket loop

Lateral Pocket loop

Twin Loop

Accidentals

Arch

An arch is the simplest pattern referred to as pattern less configuration. They are characterized by slight rise (elevation) in the ridges, which enter on one side of finger print pattern and exit on the opposite side. The Arch is of two subtypes:

Plain Arch (A)

It is the simple of all fingerprint patterns. In plain arch ridges enter on one side of impression and flow or tend to flow out on the other side with a slight rise or wave in centre. It has no Triradius and core.

Tented Arch (T)

It is the one in which most of ridges enter on one side and flow upon the other side making the sufficient recurve. Tented arch appears to have triradius near the mid axis of the finger towards the proximal end. The erect radiant is associated with abrupt elevation of transversely coursing ridges forming the "tent" which give the name to pattern.

Loop

It is one of the most common patterns found in fingerprint. A Loop is that type of fingerprint pattern in which one or more ridge enter on either side of the impression, recurve, touch or pass on imaginary lines drawn from delta to core terminate or lend to terminate towards the same side where such ridge entered. It has only one delta. Loops are sub-divided into two main types:

Radial Loop (RL)

It is so called because ridge flow or terminate in the direction of radius bone of forearm. In case of right hand finger, ridges slant toward left and in left hand finger, the slant is toward right side.

Ulnar Loop (UL)

It is so called because the ridges flow or terminate in the direction of ulna bone of the forearm. In case of right hand finger, the ridges slant toward right side and for left hand finger, ridges slant towards left side.

Whorl (W)

A whorl is characterized by a circular pattern having one or more ridges revolve around the core making a complete circle, at least two deltas are present with in a recurve in front of each whorl type. It is the most complex of all the three types. According to Henry there are two kinds of whorl i.e. true whorl & composite whorl.

Plain/True Whorl

True whorl possesses two Triradius and at least one ridge making a complete circuit which may be spiral, oval, circular or any variant of the circle. A frequent configuration is a succession of rings or eclipse. If this ring happens to be concentric then such a whorl is called as whorl concentric circle. Another common arrangement is spiral course around the core is either clockwise or anticlockwise directions. This pattern is called spiral whorl when the ridges form one spiral around the core it is called single spiral. If there are two spirals distinct in nature with two different cores, it is called as whorl double spiral

Composite Pattern

Composite patterns are compound pattern in which two or more designs each

conforming to general aspect of one of the simpler types combined in pattern area. Two or more triradius are present. There are four main type of composite recognized:

Central Pocket Loop

Central pocket loop is a composite pattern in which most of ridges take the form of a loop. It is essentially a whorl of reduced size lying in the interior of pattern area, which is constructed mainly as a loop the central pocket loop has two deltas. It is a pattern intermediate between a whorl and pure loop.

Lateral pocket Loop

In lateral pocket loop one loop serves as a side pocket to other pocket to other loop. The pocket is formed by downward bending on one side of ridge of other loop before they recurve. The ridges about the centre, the lines containing the point of core of loops have their exit on same side of delta.

Twin Loop

In twin loop there are two distinct loops, one resting upon or encircling the other and the ridges containing the point of core have their exit toward different deltas. According to United States Federal Bureau of Investigation expert, the lateral pocket loop and twinned loop should be consolidated under double loop pattern because of the complexity involved in locating and tracing the loops.

Accidentals

Accidentals are certain composite patterns within the whorl group that occur rarely and formulated purely by chance. Accidental whorl is a pattern consisting of combination of two different types of patterns excepting plain arch possessing two or more delta formation like whorl and loop, tented arch and loop triple, loops and other bizarre configuration not original to the standard type.

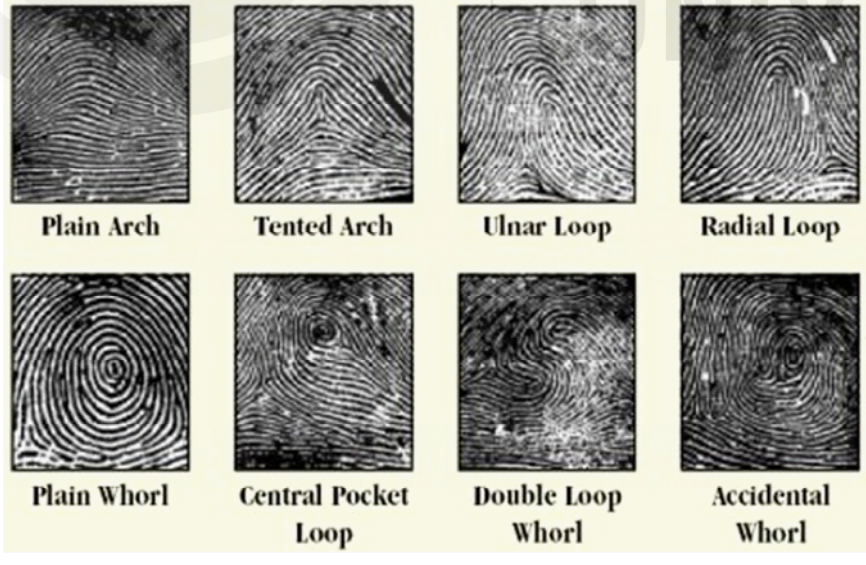

Fig. 1.7: Different types of Finger Dermatoglyphics
(Source: www.viewzone.com)

Distribution: It has been observed that among the Indian populations the frequency of loops is higher (53 per cent) than whorls (44 per cent) whereas the

frequency of arches is 3 per cent. When literature on the distribution of finger dermatoglyphics is examined in different zones, high frequency of whorls was observed from the population of Islands (50 per cent) followed by central (47 per cent), east (45 per cent), north (43 per cent), south (42 per cent) and west (39 per cent) India and the frequency of arches varies from 2 and 4 per cent in these zones (*Bhasin*, 2009).Among the tribal population the frequency of whorls was found to be highest (60%) followed by the loops and the frequency of arches was approximately 8%. The frequency of whorls among different zones was found to be highest among east India followed by central and north India followed by west and south India. phenylthiocarbamide (PTC) tasting ability.

Apart from the above, the other genetic markers such as phenylthiocarbamide (PTC) tasting abilityand colour blindness; biochemical markers like serum proteins (eg. Haptoglobins (Hp) and Transferrin (Tf)) and Red Cell Enzyme markers (eg. Glucose 6-phosphate Dehydrogenase (G-6-PD), Acid Phosphatase (ACP) and 6-phosphogluconate Dehydrogenase (PGD) etc., are also widely used by anthropologists to understand population diversity.

1.3.4 Molecular Markers

Molecular markers such as mitochondrial DNA(mtDNA), Y chromosomal and autosomal markers of chromosomal DNA are used in studying the population variation in India. mtDNA is inherited from mother while Y chromosome is contributed by father to his children whereas autosomes are contributed by both parents. mtDNA and Y chromosome studies inform about uniparental and gender based origin, history, migration and evolutionary relationships whereas autosomes represent genomic information and enlighten on prehistory and genetic admixture among populations. mtDNA studies employing hypervariable region I and II, Intergenic COII/ tRNA (Lys) 9 bp deletion polymorphism, haplotypes and haplogroups revealed that most of the populations share M haplogroup and U haplogroup was contributed by west Eurasians. Correlation of mtDNA distances of castes with social ranking was observed. Austro-Asiatic speakers were found to be original inhabitants of India. Unique haplogroups such as M31 and M32 in Onge and great Andamanese were discovered and single matrilineage of castes and tribes of India was observed. Y chromosomal studies used markers such as single nucleotide polymorphisms (SNPs), short tandem repeats, haplotypes and haplogroups in Indian population and found a distinct haplogroup variation between castes and tribes; recent admixture of Indo-European speakers into the Indian population; establishment of genetic linkage of Indians with Australians during Holocene; and genetic homogeneity of Indian Muslims with non-Muslim neighbours. Autosomal data on Indian populations was mainly comprised of SNPs, short tandem repeats, insertion/ deletion polymorphism, haplotypes and haplogroups. In autosomal studies, genetic affinities between populations were investigated. Genetic affinity was found among geographically close populations. Intermediate status of Indian populations in the genetic closeness between east and west Asian populations was observed. Genetic affinity of high caste populations with Central Asians and formation of ancestral north (ANI) and south Indian populations (ASI) by ancient human immigrants to India through southern and northern routes, were reported. It was observed that present day Indian populations are descendants of admixture ASI and ANI populations. The former contribution was high in

south India and later to the ANI in genetic element. Within the Tibeto-Burman speakers ancestral Austro-Asiatic (AAA) and ancestral Tibeto-Burman (ATB) groups were identified. Both ASI and AAA were considered as indigenous populations of India.

1.4 SUMMARY

The primary aim of physical anthropology is to study human biological evolution and variation. In this unit we have introduced you to the terms such as population, race and ethnic group which are commonly used to categorise human groups. This unit focuses in understanding the biological diversity between races, ethnic groups and populations.For this, morphological and genetical characters have been used by biological anthropologists in understanding population diversity have been discussed.

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1.6 ANSWERS TO CHECK YOUR PROGRESS

1. Population is a group of interbreeding people but Mendelian population is a group of people who reproduce sexually or are potentially capable of doing so which focuses on the evolutionary feature.

2. One of the morphological characters that have been used in studying population variation is epicanthic fold. This structure is due to a fold in the eyelid. Its appearance is also due to differences in general facial structure such as size of the cheekbones and the amount and distribution of subcutaneous fat in the face.

3. Biological anthropologists primarily use stature (height vertex), Cephalic Index and Nasal index in population variation studies.

4. Among the Indian populations the frequency of allele O is more than 50% which is followed by B and A.

2.0 SOURCES OF GENETIC VARIATION*

Contents

2.0 Introductiohn

2.1 Mendelian Mutation

2.2 Genetic Recombination

2.2.1 Crossing Over

2.3 Gene Flow and Genetic Drift

2.3.1 Gene Flow: Concept

2.3.2 Genetic Drift: Concept

2.3.2.1 Bottleneck Effect

2.3.2.2 Founder Effect

2.4 Selection

2.5 Summary

2.6 References

2.7 Answers to Check Your Progress

Learning Objectives

After reading this Unit, you would be able to:

- Elucidate various sources of genetic variation;
- Discuss Mendelian Mutation and Genetic Recombination; and
- Explain the concepts of Genetic Drift and Selection.

2.0 INTRODUCTION

Understanding biological diversity is a constant effort of Biological Anthropologists for so many decades. In the area of anthropological genetics, population genetics always provide a significant contribution to enrich the overall concept. Mendel's Law of inheritance showed us the pathway of understanding inheritance pattern and the reason behind the process. The process of selection and transmission of the trait through different generations explain the law intimately. To create the genetically new population, combination and recombination are always tested and selected by the nature contrasting its ancestral composition of population structure. Therefore, it is important to understand the reason behind these shuffling of alleles within the genetic environment. At the population level, gene flow and genetic drift tells us the story of genetic changes. Mutation to Selection – a long journey has been travelled by the alleles through a bottle necking effect for unleashing the new combination of allele by expressing new genetic traits to the nature.

Here we are going to discuss, each and every characters of the genetic play of individual – population – environment relationship. The unit 'sources of genetic variation' has been divided into five concepts as Mendelian Mutation, Genetic Recombination, Gene Flow, Genetic Drift and Selection.

*Contributed by Sri. Suvendu Maji, Department of Anthropology, University of Calcutta, Kolkata.

2.1 MENDELIAN MUTATION

Before initiating the courseware regarding Mendelian Mutation, we have to know, who was Mendel? As all of you know from your School days, Gregor Johann Mendel, popularly known as Mendel, positioned in the scientific community as father of Genetics. He did various experiments choosing garden pea plant, Pisum sativum which offered clear advantages for the genetic investigation. Now what was that genetic investigation? Mendel wanted to study heredity with 34 varieties of peas and spent about two years to select the few varieties for using them to his experiments. He verified that each variety was genetically pure by growing the plants for two generations and confirming that all offspring were the same as their parents. Here all the varieties were homozygous for each of the traits that was chosen for the study. He then carried out a number of crosses between the different varieties. Mendel conducted crosses between different plants by opening the buds before the anthers were fully developed, removing the anthers, and then dusting the stigma with pollen from a different plant. Then he began by studying monohybrid crosses— those between parents that differed in a single characteristic. Based on the results by monohybrid crosses among pea plant, he understood the common morphological traits distributed among the off-spring generation have a unique pattern. Here we must introduce the terms Phenotype and Genotype to explain the genetic inheritance pattern. All the common characters, like round shaped peas are dominantly more distributed than the wrinkled one. As we know the fundamental concept and nature of the Mendelian experiment on Pea plants from school level, the present course may be grounded on your past knowledge. Still for your quick recap, here is the three different laws of the Mendel after observing the cross results of the pea plants.

Law	Definition
Law of segregation	During gamete formation, the alleles for each gene segregate from each other so that each gamete carries only one allele for each gene.
Law of independent assortment	Genes of different traits can segregate independently during the formation of gametes.
Law of dominance	Some alleles are dominant while others are recessive; an organism with at least one dominant allele will display the effect of the dominant allele.

The word ‘segregation’ explains the nature of distribution of the genetic traits among the offspring generation (F1) from parental generation (P). The content of ‘independent assortment’ illustrates the structure of segregation. Whereas the word ‘dominance’ explains the most commonly observed traits among assorted peas. Mendel has conducted two levels of the experiments based on the selected peas. The monohybrid cross and dihybrid cross experiments based on the different variety of morphological characters. Like the Mendel’s experimental outcome, biologists explored similar fashion of inheritance pattern in animal and human species as well. As the present content deals with Human inheritance pattern that follows Mendelian properties of inheritance, we would like to explain further based on the human characters only.

Before initiating the next phase of elaboration, let us quickly revise few operational and basic definition of key terms such as, allele, loci, mutation, dominant, recessive etc.

Allele and Allele frequency: Allele is an alternative form of gene and allele frequency may be understood as the distribution of any allelic variants among a specified population.

Loci: In singular, known as locus. It is the specific chromosomal position of a specific allele or gene.

Mutation: It is a subtle and specific permanent change at chromosomal or gene level. Mutation means any heritable change in a gene. The Watson-Crick structure of DNA also suggested that, chemically speaking, a mutation is a change in the sequence of bases along the DNA. The change may be simple,such as the substitution of one pair of bases in a duplex molecule for a different pair of bases. For example, an A—T pair in a duplex molecule may mutate to either T—A, C—G, or G—C.

Dominance and Recessive: These are two different set of expressions of a specific allele. Morphologically the most common expressive characters will be dominant over the less distributed genetic traits. The mode of expression depends on the nature and composition of the allele. In simple dominant inheritance, a trait never skips a generation. Whereas recessive expression is very much specific to generation and somewhat sex related.

A single recessive allele will not affect the phenotype of a person; only when both of a pair (dd) is present is there any effect. Therefore, both parents of children with a recessive trait must carry at least one specific recessive allele.

Phenotype and Genotype: At least, in Mendelian inheritance pattern, one may explain Phenotype is the outcome of a genotype. The expression of a gene is the phenotype of its genetic counterpart.

Now let us start to understand the concept of Mendelian Mutation. Why a mutation is called Mendelian mutation?

As we know, Mutation is certain changes within any chromosomal part or in any genetic sequence. Chromosomal mutation results structural changes within our genetic structure, whereas gene mutation is very much point based mutation. It alters the functions of a gene at its expression level.

AT ATC ATAGTAGTCAGTC~~ATAAG~~TAGT AGT CAGTC

In the above hypothetical genetic sequence diagram, the TCA has replaced by AGT is an example of genetic mutation. Now hypothetically it may alter the function of the expression related to this sequence only. Now one may argue what is structural changes in this level. As we have learnt the Chromosomal mutation before, the structure changes or aberrations may be identified by this type of mutation. In such cases, we have learnt in basic mutation chapter, regarding the process of inversion, duplication, deletion and translocation within the chromosomal part.These mutations are uncertain and accidental. Although biologists have studied the pattern of these mutations that result deficiencies within any genetic environment. People have understood the

structure and function of these mutations based on different genetic disease and syndrome. The distribution pattern of the deficiencies among human families tells us the story of inheritance pattern of the particular genetic abnormalities or deficiencies. After having a critical review on the distribution pattern of any genetic abnormalities, we are referring the Mendelian inheritance pattern for identification of the disease under study. Based on the identification of studied disease inheritance pattern, we generally classify and identify the disease under any of the four Mendelian inheritance patterns. In case of studying Mendelism in man, we generally follow Autosomal dominant, Autosomal recessive, Sex Chromosomal dominant and Sex Chromosomal recessive inheritance pattern. Any chromosomal aberrations or genetic mutations, that lead to any Syndrome or genetic disorder in human may be studied referring the mendelian laws. In human, we found different diseases follow Mendelian principle especially in case of syndrome. The product of genetic mutations, that follow the Mendelian inheritance pattern may be understood as Mendelian Mutation. This is worth mentioning that these genetic disorders that follow Mendelian Law of inheritance are based on primarily single gene, known as single gene disorder.

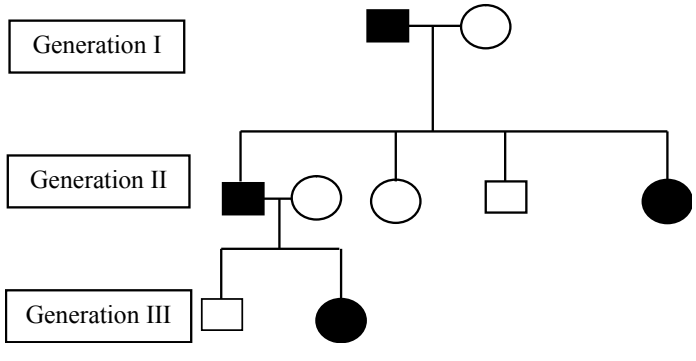
To understand the Mendelian Inheritance pattern, we have to study the basic principle of the four different inheritance patterns. In human genetics we may understand such issues based on family study by analyzing Pedigree or popularly known as family tree. In case of genetic counselling, this is the first step to understand the genetic background of the disease within family members.

Features of Autosomal Dominant Inheritance:

- All the affected individuals have at least one affected parent. Also affected individuals are usually found in every generation of a sizable pedigree.
- Males and females are affected in about equal numbers, which makes X linkage less likely and rules out Y linkage.
- Both males and females transmit the trait.
- Mating between heterozygous affected with unaffected individuals produce about ½ affected and ½ unaffected offspring.
- The trait does not appear in the descendent of two unaffected parents.

Few examples – Familial Hypercholesterolemia, Huntington disease, Marfan syndrome, Myotonic dystrophy, Neurofibromatosis, Retinoblastoma, Achondroplasia or Dwarfism etc.

Hypothetical pedigree explains the mode of inheritance pattern of Autosomal dominant trait:



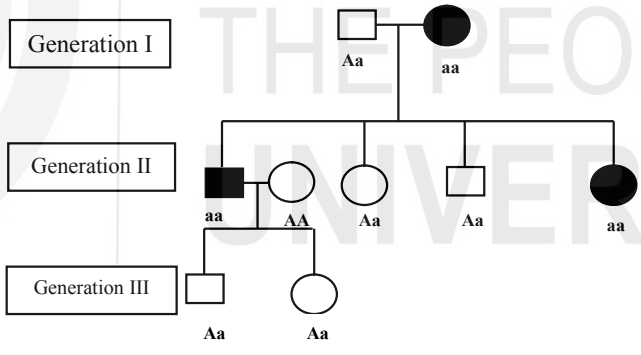
Features of Autosomal Recessive Inheritance:

- Most affected individuals have two unaffected parents.
- The trait is expressed in both the sexes and transmitted by either sex to both male and female offspring in roughly equal numbers.
- Mating between unaffected heterozygotes yield about ¾ unaffected and ¼ affected offspring.
- Mating between an affected and an unaffected parent will usually yield all unaffected offspring; only if the unaffected parent is heterozygous can affected children be produced.
- When both the parents are affected, all their children are affected.

Few Examples: Cystic Fibrosis, Sickle cell disease, Albinism, Galactosemia, Gaucher disease, Hurler syndrome, Tay-Sachs disease, Thalassemia (α and β) etc.

In this pattern, we will gradually understand the concept of carrier. If the disease carried by heterozygous condition where the expression of the disease will not be expressed phenotypically, though the genotype holds half of the dosage of diseased allele for a generation. It will express only when half of its abnormal dosages of allele complete the recessive composition of the allele which is rare.

Hypothetical pedigree explains the mode of inheritance pattern of Autosomal recessive trait:

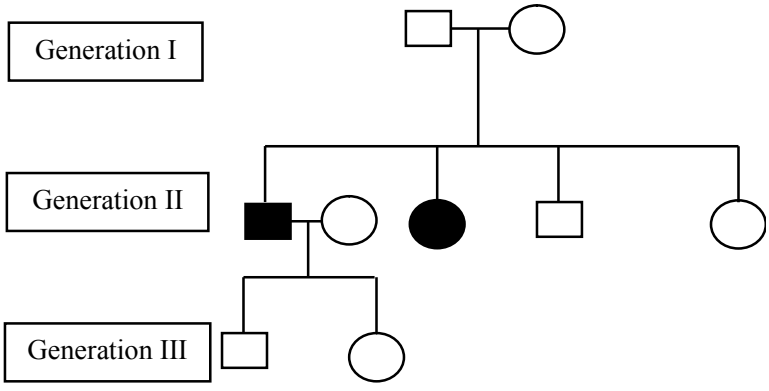


Features of Sex Chromosomal or X- Chromosomal Dominant Inheritance:

As this pattern follow the trail of X-chromosome, no male-to-male transmission of the trait.

- Affected males when mated to unaffected females produce all unaffected sons and all affected daughters.
- Affected females can produce affected sons and daughters as well as unaffected sons and daughters. The ratio of affected to unaffected is about 1.1.
- Every affected individual has one affected parent; but for affected males, the affected parent is always the mother.
- Males, being hemizygous, are usually more severely affected than females, who are heterozygous.

Examples: Hypophosphatemia, also known as vitamin D-resistant rickets.

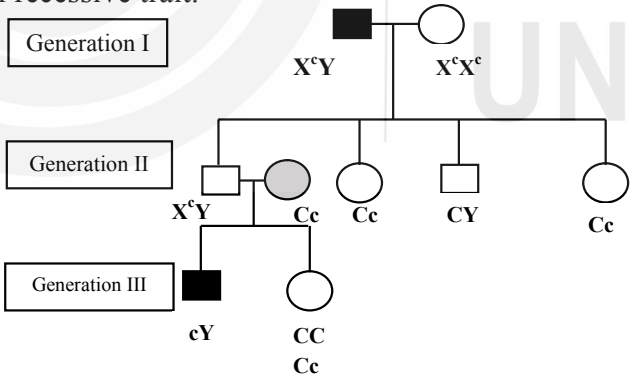


Features of Sex Chromosomal or X- Chromosomal Recessive Inheritance:

- Father-to-son inheritance is never seen. Instead affected males always get their abnormal allele from their mothers. Male individuals are affected more than female individuals in a family.
- Among the sons of carrier mothers, about half are affected and half are unaffected.
- All the daughters from the mating of a carrier female with an unaffected male are unaffected, but half will be carriers.
- The mating of an affected male with a homozygous unaffected female produces no affected offspring, but all their daughters will be carriers.

Examples: Duchene Muscular Dystrophy, Hemophilia, G6PD deficiency, Hunter syndrome, Lesch-Nyhan syndrome etc.

Hypothetical pedigree explains the mode of inheritance pattern of Sex Chromosomal recessive trait.



2.2 GENETIC RECOMBINATION

If we focus on to analyze the unit "Sources of Genetic Variation", we understand that the word 'variation' indeed holds the important aspect of the present objective of study. Studying 'variation' is one of the central themes of Anthropological Studies. Like other branches of Anthropology, Biological Anthropology contributes a lot of information regarding Human Biological variation through classical genetic markers, biochemical traits to molecular markers in one hand and Anthropometry in other. To understand the source of biological variation, we need to understand the concept of recombination at genetic level.

Recombination can be simply defined as the process by which chromosomes exchange genetic material. In Meiosis, new combinations of the parental genetic material are created by transmitted to offspring. The process of recombination among the different alleles at the time of transmission from parental generation to offspring generation is well understood at cell biology level but still we don't have any technology discovered till now by which we may control the composition of the allele at the time of cellular fission-fusion.

Let us narrate a situation where you have to choose a room mate between two of your best school friends based on the distance of residence. Your teacher told you to pick up one name from a student list among them. Now there is a dilemma going on to your mind. You chose friend A, though B will be more suitable for you. Why you chose A instead of B? May be the distance of the residence between both of you are closer than the other one. Like this, the half of Allele A don't know how to choose other half its body. Nature told them to choose based on the distance from each other. The distance between two loci will be the determinant factor behind these permutation and combination. The map distance between two loci is known as Centimorgan (cM). The loci which are situated within 1 cM are more likely to be linked with the others. Theoretically, loci situated within 1cM will behave as linked, whereas more than 1 cM-based loci will be accounted for more variation by crossing over between two alleles.

So, now we have to understand Crossing Over (CO) for comprehending human genetic variation.

2.2.1 Crossing Over

In Cell Biology, we have learnt Cell division through Mitosis and Meiosis. Crossing-over is the primary means by which the disjunction of homologous chromosomes at meiosis I (MI) is ensured. Recombination events themselves do not commit chromosomes to segregate; rather, disjunction is mediated by specific structures, known as chiasmata, which are formed at the sites of exchange. This is a process by which homologous segments of paired maternal and paternal chromosomes are exchanged. Its effect is that even genes that are "linked" in a single chromosome do not necessarily remain together from one generation to another, but are interchanged with their alleles in the homologous partner chromosome. Because Meiosis assorts and recombines the genetic contributions of an individual's parents, it has been compared to the shuffling of cards.Depending upon the number of chiasmata appearing in the chromosome, we have Single, Double and Multiple type of Crossing Over. In brief, crossing over is the process of exchange of genetic material or segments between non-sister chromatids of two homologous chromosomes. It occurs due to the interchange of sections of homologous chromosomes. This is the most important phenomena behind the enormous variation found in nature from single cellular organisms to human being.

Centrimorgan (cM):It is a map unit used to express the distance between two gene loci on a chromosome. A spacing of one centimorgan indicates a one per cent chance that two genes will be separated by crossing over. One cM is equal to a 1% chance that a marker at one genetic locus will be separated from a marker at another locus due to crossing over in a single

generation. In humans, 1 cM is equivalent, on average, to 1 million base pairs. The centimorgan is named after the pioneering (and Nobel Prize winning) geneticist Thomas Hunt Morgan. (URL: https://www.medicinenet.com/script/main/art.asp?articlekey=2665)

Check Your Progress

1) Name one autosomal dominant and one recessive disorder.

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2) What causes recombination?

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3) Is genetic recombination the same as crossing over?

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2.3 GENE FLOW AND GENETIC DRIFT

2.3.1 Gene Flow: Concept

The term 'Gene flow' is quite popular in the study of population genetics. As the term itself explains the inclination towards mobility and migration of a group or individual to a new space. When an individual migrates from one population to another, it carries genes that are representative of its own ancestral population into the recipient population. If the individual successfully selected into the new environment and start breeding, it will transmit those genes between the populations.Now the process of transfer of genes is known as gene flow. So, from the population genetic perspective, we cannot use the term migration or gene flow as synonymy. Critically we may state that all the gene flow indicates migration, but all the migrations are not completing the process of gene flow until it breeds with the recipient population.

Generally, proximally placed populations are more likely to choose one another as breeding partners. In some case of Human population, marriages will be preferred between closely situated population and on the basis of social identity. Although, theoretically Human population are not behaving like other animal species where geography may influence as a determinant factor for the creation of new gene pool. In this scenario, 'reproductive isolation' as defined in the biological species concept can be viewed as a kind of inverse of gene flow. Religion, language and other cultural attributes play constant role in reproductive isolation in humans. Think about India, this is the best example

of the different size of diverse population holding unique gene pool. From the isolated or connected geographical space, small to large ethic communities, different caste groups, number of different linguistic and religious groups, it is a product of preferential marriage system coming from time immemorial to this land of diversity. Due to a large number of chaotic movements within, between and among these groups, India becomes a unique example of human biological diversity. To maintain these diversities, gene flow acts as an important role to shape the population structure of the recent time. Gene flow acts as one of the important evolutionary forces to maintain the genetic stability by altering allele frequency within a population. Gene flow between the reproductively isolated gene pool will produce more heterogeneity through number of generations. Reproductive isolated mechanism behaves as a barrier to gene flow. In case of human population, immigration shares a lot of new genes to the recipient population. The new breeding population will produce diversity that cater new combinations of allele for stabilize population.

Gene Flow or gene migration, takes place when individuals migrate from one population to another and interbreed with its members. Gene frequencies are not changed for the species as a whole, but they change locally whenever different populations have different allele frequencies. In general, the greater the difference in allele frequencies between the resident and the migrant individuals, and the larger the number of migrants, the greater effect the migrants have in changing the genetic constitution of the resident population.

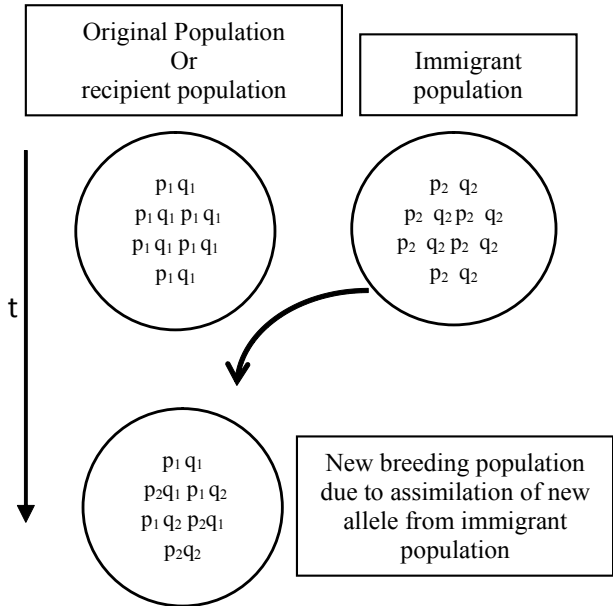
(Ref:https://www.britannica.com/science/evolution-scientific-theory/The-science-of-evolution#ref311620)

In case of Human Population, two genetically different, geographically distantly placed groups are interconnected by a series of populations of intermediate type through which genes can flow and give rise to geographical gradient of gene frequency between two distant populations. For example, the gradual reduction in the frequency of gene B for blood group B from higher value in Central Asia to its lowest in western Europe i.e. there is a decrease in gene B from East to West. This largely affected by the repeated invasion from the east to which Europe has been exposed.When this type of gene flow happens in one direction from local population then it decreases frequency along a line called cline.

Now we may move towards little bit technical aspect of gene flow. As we have learnt that gene flow alters the allele frequency of recipient population where the genes are getting assimilated. The change in frequency of a gene in a population following the introduction of individuals from any other population depends upon the number of immigrants and the frequency of gene in question among them. If the frequency of two alleles A and B is p1 and q1 in the original population respectively and p2 and q2 among the immigrants.

Original Population: allele A – allele frequency p1 and allele B – allele frequency q1

Immigrant population: allele A – allele frequency p2 and allele B – allele frequency q2



The new breeding population made up of a fraction X of the original population and the fraction Y for the immigrants. Now the frequency of A becomes Xp1 + Yp2 and B becomes Xq1 + Yq2. This is a simple hypothetical explanation of gene flow. Now you understood the fundamental concept of gene flow and why it is important to study related to sources of human variation.

2.3.2 Genetic Drift: Concept

Now we have learnt Gene flow, we have understood that new combination of alleles come into existence into the environment due to gene flow. Now we have to understand how gene flow bring changes into a recipient population. This is little bit technical part of this section of study. The process by which the allele frequency change into a breeding population is genetic drift. Genetic drift refers to random fluctuation of allele frequency by chance. The effect of genetic drift is usually the most recognizable in small population.

Genetic drift can lead to disappearance of an allele from a population or it may also lead to a higher frequency of an allele in the same population. It means that when new alleles get a chance to breed with the allele of recipient population, it creates new combination of alleles. If the new combination of alleles gets selected into the environment, it may frequently increase its number compared to other combinations of allele. Sometimes due to selective advantages of the new combinations of allele over power the rare alleles gradually get disappeared from the same environment. When a population is affected by genetic drift, the frequency of an allele will randomly change from one generation to the next generation until the allele is either fixed or lost. In a small population it may lead to extinction of a population or the population may well adapt to the environment and become widely divergent from the parental population. In due course it may lead to origin of new species especially in island or reproductively isolated population.

- ❖ Genetic drift is a random process; Random events play a strong role in evolution, especially in small populations.
- ❖ Natural selection and genetic drift both result in a change in the frequency of alleles in a population, so both are mechanisms of evolution. Natural selection is not a random process.

❖ One of the requirements for the maintenance of allele frequencies in populations should be of large population size.

Genetic drift

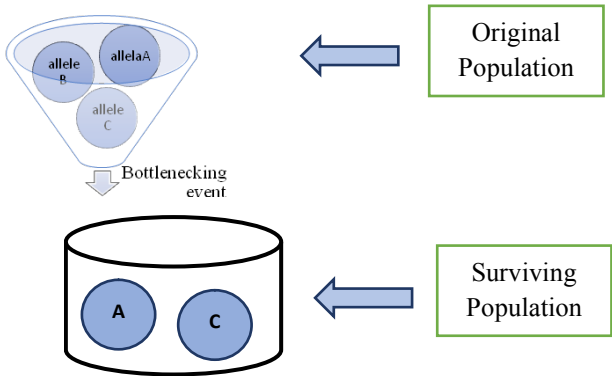
1) Genetic drift is a random process.
2) It alters the allelic frequency at genotype level.
3) Due to bottlenecking process survival population possess selective alleles.
4) Due to founder effect new set of alleles produce new gene pool.
5) It is observed evidently in the small population.

Before explaining the other facet of this section, let us understand, what is fixation? Let me give you an example of two alleles A and B where p and q are the frequency of A and B alleles of a population. If we think the allele is get selected and commonly distributed within the environment then, the allele frequency will be 1.00. Here the fixation is defined as allele frequency reaching p=1 or q=1. Allele frequency is represented by the following equation p + q =1 where p is the allele frequency of dominant allele and q is the allele frequency of recessive allele at a given locus and the sum of these two equals to 1 or 100 %. In population genetics, we have studied the concept of Hardy-Weinberg equilibrium. Here the equilibrium means balance, theoretically the perfect balance estimates as 1.When we are going to estimate whether the population is under Hardy-Weinberg equilibrium, we usually check, the aggregate of all the allelic frequency that should tends to 1. Suppose there is a population of 1,000 individuals out of which 900 are of blood group O and the rest 100 are of blood group B. Several factors for instance illness, accident, disaster etc. may gradually eliminate individuals having B blood group since they are less in number. Thus, in this context increase or decrease of a particular allele does not depend whether an allele is advantageous or disadvantageous but happens merely by accident or by chance. Therefore, sometimes it is referred to as chance fluctuation or accidental loss or fixation of a gene. Here we try to understand whether the population experienced the influence of the genetic drift through fluctuation then we have to estimate the proportional changes of allele frequency of A and B blood group into a population. The population always tries to adapt new strategies through different selective mechanism by changing the allele frequency and try to maintain the balance tends to 1. Genetic drift is powerful when it is not opposed by selection: that is, when drift is between different neutral forms. However, in Wright's theory, drift has to work in opposition to selection. Genetic drift is of two forms: (a) Bottleneck effect and (b) Founder effect.

2.3.2.1 Bottleneck Effect

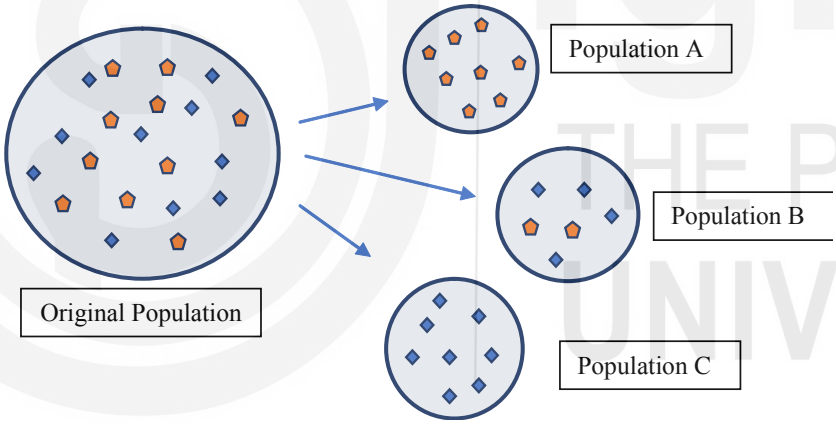
It is a form of genetic drift where there is a great fluctuation or change in allele frequency due to chance that occurred when the original population size is reduced drastically (population crash) and later the remaining members expand (population flush) by interbreeding. It is so named because the bottom of a bottle is large but it becomes smaller or narrow as it suggests the large original population is reduced in size and only a few could come

out of small neck of the bottle suggesting only few individuals are left after bottleneck effect. The enlarged population after bottleneck effect may become different from the original population in the context of allele frequency.



2.3.2.2 Founder Effect

The founder effect was proposed in 1956 by Harvard evolutionist Ernst Mayr. Founder effect is another form of genetic drift where a small group of individuals splits from the original and large population and establish a different new population. The newly formed population may not represent the total genetic diversity as in the original population i.e. the allele frequency may become different from that of original population and some alleles may be missing altogether.



The name the process, Founder Effect itself explain its underlying meaning. Here the new populations get new kinds of individuals with unique allelic combination which are not being observed to their stock population or original population. The above diagram shows equal number of diamond and pentagon individuals in the original population. The original population splits into three populations each having different number of diamond and pentagon individuals which is totally different from the original population also the small size of the new colonies means they will experience strong genetic drift for generation.

Sewall Wright is the person who had a primary contribution to explain the genetic drift in population genetics. The proper mathematical interpretation and the term "genetic drift" was coined by a founder of population genetics, Sewall Wright. His first use of the term "drift" was in 1929, though at the time he was using it in the sense of a directed process of change, or natural selection.In this area the name of Wright is regularly associated with those of Haldane and Fisher. Wright's shifting balance theory is a way of taking

advantage of gene interactions. He had long been concerned with cases in which genes interacted in ways not predictable from their individual effects.

2.4 SELECTION

Like the previous topic, selection is the most important process between biology and nature that decides the changes will remain at genetic environment or reject from the environment. Darwinian concept of natural selection explains the numerical size of a population may help them to struggle more for their survival. Phenotypic variation exists among individuals and the variation is heritable. Individuals who are reproductively fit and inherit selective heritable traits are more likely fit to the environment. Darwin's theory consisted of two main points; 1) diverse groups of animals evolve from one or a few common ancestors; 2) the mechanism by which this evolution takes place is natural selection. Darwinian selection refers natural selection though the finer differences between two process depends at the individual level and population level. In case of Human population, sexual selection is not random and therefore the effect of the non-random breeding population yields specific selective phenotypes. Natural selection is also limited here because it acts on the phenotypes of individuals, not alleles.Any given individual may carry some beneficial alleles and some unfavorable alleles. Natural selection acts on the net effect of these alleles and corresponding fitness of the phenotype. As a result, good alleles can be lost if they are carried by individuals that also have several overwhelmingly bad alleles; similarly, bad alleles can be kept if they are carried by individuals that have enough good alleles to result in an overall fitness benefit. In case of human population, due to preferential marriage system of the society aggravates these types of gain or loss of good alleles. Theoretically the natural selection process demands large numerically large number of finite breeding population that may randomly choose their sexual selection. There are five types of selection that may affect population variation. The five different types of selection are as follows:

- stabilizing selection
- directional selection
- diversifying selection
- frequency-dependent selection
- sexual selection

As natural selection influences the allele frequencies in a population, individuals can either become more or less genetically similar and the phenotypes displayed can become more similar or different as per their genotypic composition. In the end, natural selection cannot produce perfect organisms from its origin, it can only generate populations that are better adapted to survive and successfully reproduce in their environments.

The assumption of basic Selection Model are as follows:

Genetic System

1) Single, Biallelic, Autosomal locus
2) Diploid
3) Random Mating among Individuals

Selection

1) Selection identical in both sexes
2) Selection occurs through differences in viability
3) Selection constant in space and time for each genotype

Other factors

1) Non-overlapping generation
2) No mutation
3) Large population size
4) No gene flow
5) No inbreeding

In case of stabilizing selection, natural selection favors an average phenotype by selecting against extreme variation, the population will undergo at a stabilized condition. When the condition is getting stabilized, the chances of fluctuation through different evolutionary forces will be decreased as well. This state of affair diminishes the diversity of a population and gradually new combinations of allele through new phenotypic expression will be less. For example, Human birth weight is a polygenetic selection which is controlled by environmental factors. Infants with low birth weight are weak in underdeveloped countries and experience health problems, while large babies of developed countries will have problems passing through the birth canal. Whereas babies with average birth weight are more likely to survive than a too small or large baby. The intensity of that selection has decreased as medicine has improved for small or too large babies by a few weeks in an incubator or delivery by caesarian section.In case of directional selection, the environment changes, which selects for phenotypes at one end of the spectrum of existing variation. The pattern of allelic selection through different phenotypic advantages especially in human population, steadily incline towards some specific kinds of allelic recombination. The result of this type of selection is a shift in the population's genetic variance toward the new, fit phenotype. For example, nature is not kind to white lemurs. The directional selection could exist in a single source, such as a new predator that only eats white lemurs. For example, in addition to a new predator, white lemurs could also be subject to increased sunburns, which might decrease their reproductive success. Diversifying (or Disruptive) selection can also occur when environmental changes favor individuals on either end of the phenotypic spectrum. Sometimes natural selection can select for two or more distinct phenotypes that each have their advantages. The intermediate (phenotypes) individuals are often less fit than their extreme counterparts. Imagine a population of mice living at the beach where the space is full of light-colored sand interspersed with patches of tall grass. In this situation, the light

and dark colored mice would be favored in sand and tall grass respectively. Whereas the medium colored mice would be more vulnerable and probably be eaten by predators due to its medium colored skin color. In case of sexual selection of human population, the selection pressure on males and females for their marriages influence the random nature of sexual selection processes. The sexual selection has its two different forms: intersexual selection (also known as 'mate choice' or 'female choice') in which males compete with each other to be chosen by females; and intrasexual selection (also known as 'male–male competition') in which members of the less limited sex (typically males) compete aggressively among themselves for access to the limiting sex. For example, in Non-human primates, Male baboons (Papio) are more than twice as large as females, and the behavior of the docile females contrasts with that of the aggressive males. The larger male Baboons usually enjoy a hierarchy of dominance based on age and strength, with males that rank high in the hierarchy doing most of the mating.

Check Your Progress

4) What is bottleneck effect?

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5) What are different types of selection?

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2.5 SUMMARY

So, now we may summarize the entire portion of the Unit 'Sources of Genetic Variation' that consists of Mendelian Mutation, Genetic Recombination, Gene Flow and Genetic Drift, Selection. We have started with Mendelian Mutation and Genetic Recombination. These two sections dealt with theory of the different processes of genetic changes. As Mendelian contribution is important to understand the mutation at genetic level and its processes as well. We have revised the law of inheritance by Mendel and then learnt the different inheritance pattern in human or we may say Mendelism in man. Genetic recombination explains the gene level mechanism for the creation of variation in nature. Here, we have studied Linkage and Crossing over as the most important factors for genetic recombination. Then Gene flow and drift section narrated the story behind the changes in population structure from population genetic perspective. At the end of the unit, Selection process was explained to understand the relationship between nature and individual.Sources of genetic variation explain all the fundamental facets to understand the scientific background of the changes in population.

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2.7 ANSWERS TO CHECK YOUR PROGRESS

1) Autosomal Dominant: Huntington disease and Autosomal Recessive: Sickle Cell Disease.

2) Genetic recombination is caused by chromosomal exchange of genetic material. For details refer section 2.2.

3) Recombination is the process by which chromosomes exchange genetic material whereas Cross-over is the primary means by which the disjunction of homologous chromosomes at meiosis I (MI) is ensured. For details refer section 2.2.

4) Bottleneck effect is a form of genetic drift where there is a great fluctuation or change in allele frequency due to chance that occurred when the original population size is reduced drastically (population crash) and later the remaining members expand (population flush) by interbreeding. For details refer section 2.3.

5) There are five different types of selection: stabilizing selection, directional selection, diversifying selection, frequency-dependent selection and sexual selection.

UNIT 3 GENETIC POLYMORPHISM*

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3.8 Answers to Check Your Progress

*Contributed by Dr. SAA Latheef, Department of Genetics and Biotechnology, Osmania University, Hyderabad.

Learning Objectives

After reading this Unit, you will be able to:

- Understand the distribution of genetic markers in Indian populations;
- Elucidate the causes and applications of genetic variations in humans;
- Discuss the polymorphisms in blood groups, serum proteins and red cell enzyme/proteins; and
- Understand parental affinities of Indian population with respect to the distribution of mtDNA, Y chromosome and autosomal markers.

3.0 INTRODUCTION

India is ranked 6th in geography, second populous country, has 1.3 billion populations (world meters.info), occupy 31st position in population density and one by sixth in world population. The population is distributed in 28 States and 8 Union territories. The per capita income of Indian population is 8378 dollars. On the basis of climate, India is divided into eight types namely tropical, savannah, monsoon with short dry season, monsoon with dry season in high sun period, semiarid and steppe, hot desert, monsoon with dry winter, cold humid winter with short summer and polar (Bhasin, 2009). Politically, India is segmented into six regions (north, south, central, west, east and islands). As per census of India (2011), the sex ratio was 940 females per 1000 males, had literacy rate of 74%, composed of 4693 communities, eight religions (Hinduism, Islam, Sikhism, Christianity, Buddhism, Jainism, Zoroastrianism and Judaism), 325 spoken languages spoken and 25 scripts are reported. On the basis of physical features, the Indian populations are grouped into Caucasoid, Mongoloid, Australoid and Negrito (Andaman). Malhotra (1984) observed the presence of 40,000 endogamous populations, of which approximately 37,000 are caste groups and about 3000 tribal and religious groups. Anthropological Survey of India identified 4635 communities in India. There is no consensus among anthropologists that these communities are strictly endogamous (marrying within their social group) or not.

The spoken languages in India are assigned to four language families i.e. Dravidian, Indo-European, Austro-Asiatic and Tibeto-Burman. Austro-Asiatic speakers are considered as ancient residents in India. Within each language family, populations are strictly divided hierarchically into endogamous castes, tribes and religious communities with strict rules regarding marriage. Scheduled Castes and Scheduled Tribes are specified in 15th Presidential orders empowered by Articles 341 and 343 of Indian Constitution and listed in amended Scheduled caste and Scheduled Tribes act of 1976. Tribal population of India constitute second largest indigenous population in the world forming 8.6% of Indian population numbering 705 as per Ministry of Tribal affairs, Government of India (2013). For administrative reasons, castes are divided into other castes/forward castes, backward castes, scheduled castes and scheduled tribes. In some states, economically most backward castes within backward castes are also categorized. Caste is defined as a hierarchical system of endogamous social grouping and control, where each group is assigned a ranked status depending on its origin (Reddy, 2010). Indian castes are divided

into four hierarchical groups (Brahmin, Kshatriya, Vaisya and Sudra) based on Varna system. Earlier there were occupational groups i.e. Brahmins serving as a Priests involved in religious activities, Kshatriya as warriors acted as Kings and protected Kingdoms, Vaisya as traders involved in business activities and Sudras as labourers.

Castes are subdivided into Sub Castes and Gotras, whereas tribes were sub-divided into sub-tribes (*Reddy*, 2008). Not all occupational groups are arranged into Varna and there is no unanimity on the hierarchical status of Caste and across the geography. Caste groups are immutable. Under "Jati"(smaller of smaller circles of caste) (*Bhasin*, 2006) system all caste groups are realistically arranged and mutable also. Each caste has developed rules to govern social interactions including marriage which vary across the region, language, population and kinship system.Tribe is confined to particular territory, egalitarian and self-reliant economic group whereas caste is heterogeneous social group with larger economic structure. For economic security, tribes and lower castes started imitating the beliefs, customs and life style of higher castes known as Sanskritization which resulted in acquiring the caste status to the tribe and higher caste status to the lower caste. Transformation of tribe into caste and continuum is called tribe-caste continuum.Two marriage regulations direct the levels of breeding isolation and endogamy such as endogamy and sapinda. Endogamy is marrying within own social group (example caste) which has several exogamous units which prohibit marriage within their gotras/clan. Sapinda rule impose taboo on marriage between descendants of maternal and paternal lines within five and seven generations.

Kinship system of Indian population falls into two categories namely Dravidian and Indo-Aryan.The former is found in north India, whereas the latter found in south India.In Dravidian kinship systems, consanguineous marriages, village endogamy, marriage with neighbouring villagers are preferred and they have limited marriage network and restricted number of mates while in the Indo-Aryan kinship system it is vice-versa. The Indo-Aryan kinship system is formed by single paternal lineage while the Dravidian kinship system by the multiple paternal lineages. Marriage between two families is discouraged and bride exchange is confined to one generation only in Indo-Aryan kinship system whereas exchange of brides takes place between families and marriage of two brothers with sisters of another family is not uncommon in the Dravidian kinship system. In exceptional cases in Indian population marrying of women of lower with man of higher varna (Hypergamy) is allowed with the condition that the progeny will assume the caste status of their father. Two common residence patterns are observed in India. One is patrilocal wherein the woman after marriage resides in the house of husband's and another is matrilocal residence in which husband after marriage stay with wife's family (ex. Khasi, Jaintia and Garo tribes of Meghalaya).

3.1 CAUSES AND APPLICATIONS OF GENETIC VARIATIONS IN HUMANS

Variations in humans is caused by waves of migrations and admixture with indigenous people, presence of racial elements, adaptations to the environment, isolation, mating patterns and operation of evolutionary forces such as mutations,

gene flow, genetic drift, recombination and natural selection. Study of variations are useful to investigate evolutionary relationships, evolutionary history, gender specific (mitochondrial DNA and Y chromosomes) migrations, appreciate the extent of variation in humans, enhance our knowledge on evolutionary forces, susceptibility (BRCA1 and BRCA2 genes increase the risk of breast or ovary cancer) or protection (mutations in CCR5 gene protect against HIV) to diseases and drug responses (P450CYP2CP gene variants and Warfarin drug dose requirements).

The unit of genetic variation is Mendelian or breeding population. Mendelian population is a group of sexually reproducing individuals or individuals having the potential capability of reproduction and sharing common gene pool. Indian population offer great opportunity to study variation as it is fragmented in caste, tribal, religious and linguistic groups. Though these fragmented populations live side by side for thousands of years but retain their identity by practising endogamy and also show variation in the rate of microevolution (alteration in frequency of the gene in a population over a period of time). Indian population show second highest genetic diversity after African population (Tamang and Thangaraj, 2012). As per Cavalli-Sfroza and Bodmer (1971) the occurrence in the same population of two or more alleles at one locus, each with appreciable frequency is called polymorphism. If the frequency of variation is more than 1% in particular population then it termed as polymorphism and if less than 1% it is labelled as mutation.

Physical anthropology is demarcated into old (before 1951, W. Laskar era) and new physical anthropology (after 1951-S.L.Washburn era). In Laskar's era, physical anthropologists described biological parameters (bone, teeth, body build, components of body, growth and development) to understand their causes. These parameters are still in use for exploring new dimensions. In new physical anthropology, dermatoglyphics, applications of mathematical models to understand variations, evolutionary theories, evolutionary forces, adaptation, serology, biochemical genetics, demographic studies, ecological studies, somatoscopic and somatometry studies gained currency. Human cytogenetics improved our understanding on the evolution and adaptation. Development of recombinant DNA technology, subsequent sequencing and next generation sequencing brought many tools (restricted fragment polymorphism, single nucleotide/copy number variations, microsatellites (Variable Number of Tandem Repeats and short tandem repeats), Alu insertion/ deletion polymorphism, haplotypes, haplogroups, and short tandem repeats of mitochondrial and chromosomal DNA (Y chromosome and autosomes) are extremely used markers to study human variation. In this unit, salient features on genetic markers are described from blood groups to molecular markers.

Check Your Progress

1) What are the causes and applications of variations in humans?

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3.2 BLOOD GROUPS

As of now 39 blood groups are known in humans (https://www.isbtweb.org/fileadmin/user_upload/Table_of_blood_group_systems_v6.0_6th_August_2019.pdf). In India, variation data on Indian populations with respect to blood groups is available for the following systems (ABO, MNSs, P, Rh, Lutheran, Kell, Duffy, Kidd, Diego and ABH). The allele frequencies of the above blood group systems among different Indian populations are presented (*Bhasin* 1988, 2006, 2009; *Bharti et al.*2017; *Rami Reddy*, 2017; *Ray et al*.2014; *Ghosh*, 2017).

3.2.1 ABO Blood Group System

Based on the presence and absence of antigens, ABO system is categorized into A, B, O and AB. Antigens are substances that stimulate the immune response. They are either protein, carbohydrate or glycoproteins in nature. There are two subtypes within A blood group namely A1 and A2 blood group.

ABO Blood Groups	
Blood Group A	
Allele frequency	0.18 (0.17-0.18)
Populations with high frequency	Populations of Purchaval hills, Meghalaya plateau, Eastern Himalayas, STs of southern peninsula, Mon-Khmer speakers of Austro-Asiatic, Kuki chin group of Tibeto-Chinese language families, people of Himalyan and North-East frontier group and Naga; Samanthas, Reddis, Oddes, Marines, Naik Pods, Valmikis, Rajgonds, Bagathas, Chenchus,Gadaba, Konda Doras, Parengi Porojas, Muslims of Andhra Pradesh.
Populations with low frequency	People of Assam valley, STs of western and north-eastern peninsular plateau, speakers of Indo-European and Dravidian in southern region; Nagaras, Vysyas, Devangas and Senguid-Mudaliyar, Malas, Kuruvikkarans, Yanadi and Christians of Andhra Pradesh.
A1 Blood Group	
Allele frequency	0.07-0.29
Populations with high frequency	Gond and Bhil of Madhya Pradesh; Dhodia, Gamit, Ghanchi, Vasava and Vania Soni of Gujarat; Brahmins, Jalaris, Madigas, Mala, Chenchus and Naikpods of Andhra Pradesh.
Populations with low frequency	Keer, Hindu, Muslim of Madhya Pradesh; Parsi, Kunbi, Anavil, Muslim and Kotwalia of Gujarat, Parsi of Maharashtra; Bhil of Rajasthan; Vysyas, Sengund-Mudaliyar, Konda Reddis, Jatapus, Koyas, Yanadi of Andhra Pradesh.

A2 Blood Group	
Allele frequency	0.02 (0.00-0.23)
Populations with high frequency	Dhodia and Parsi of Gujarat, Parsi of Maharashtra; Bhil and Muslim of Madhya Pradesh; Ghanchi, Kunabi, Gamit and Kotwalia, Anavil and Vasava of Gujarat; Jalaris, Mala, Naikpods of Andhra Pradesh.
Populations with low frequency	Nil in Keer of Madhya Pradesh; Mongoloid affinity, ST, Gond of Madhya Pradesh; Vania Soni and Muslim of Gujarat; Bhil of Rajasthan; Vysyas, Reddis, Kamma, Sengund-Mudaliyar, Madiga, Jatapus, Yanadi, Koya, Konda Reddi, Pradhans of Andhra Pradesh.
B Blood Group	
Allele frequency	0.23 (0.11-0.33)
Populations with high frequency	Keer, Gond, Bhil, Hindu and Muslim of Madhya Pradesh; Parsi of Maharashtra; Bhil of Rajasthan; Parsi Vania Soni, Ghanchi, Kunabi, Anavil, Kotwalia and Gamit and Muslim of Gujarat; Assam Valley, tribes of western and north-eastern peninsula; speakers of Indo-European and Dravidian in southern region; Devangas, Sengund-Mudaliyar, Mala, Kolams, Nooka Doras and Muslims of Andhra Pradesh.
Populations with low frequency	Dhodia and Vasava of Gujarat; Kammaras, Kummaris, Yerukula, Konda Kapu and Christians of Andhra Pradesh.
O Blood Group	
Allele frequency	0.58 (0.44-0.60)
Populations with high frequency	Bhil of Rajasthan, Gond, Bhil, Hindu, Muslim and Keer of Madhya Pradesh; Vania Soni, Kunabi, Anavil, Vasava, Muslim and Kotwalia of Gujarat; Vysyas, Kummaras, Nagaras, Kummaris, Konda Kapu, Yanadi, Kuruvikkarans and Christians of Andhra Pradesh.
Populations with low frequency	Ghanchi and Gamit of Gujarat; Samanthas, Mala, Nooka Doras and Muslims of Andhra Pradesh.

3.2.2 Rhesus(Rh) System

In this system, RHD (D antigen) and RHCE (CE (ce, Ce, cE, CE) Cw,Cx and VS antigens) genes encode the antigens. Red blood cells of individuals showing absence of D antigenare called as Rh negative. Three classifications for describing Rh were proposed (Fisher and Race, 1962; Weiner and Wexler, 1963 and International society of blood transfusion). For Indian population, variation in Rh system was described using Fisher and Race nomenclature known as CDE which is also popularly used for its simplicity. Various haplotypes (a set of alleles inherited together from a single parent) were described in Rh system (RH*CDe, RH*cde, RH*cDE, RH*cDe, RH*CDE, RH*Cde, RH*CdE and RH*cdE).

Rh Blood Group as per Fisher and Race,1962	
RH*D	
Allele frequency	0.80 (0.53-1.00)
Populations with high frequency	STs of India; People of Himalyan mountain complex; East India, Mongoloid affinity, tribes of western, north-eastern and southern peninsula; Gond, Keer and Bhil of Madhya Pradesh; Vania Soni, Ganchi, Kunbi, Anavil, Vasava, Kotwalia and Gamit of Gujarat.
Populations with low frequency	Absent in Bhils of Rajasthan.
***RH*d* Blood Group**	
Allele frequency	0.08-0.34
Populations with high frequency	Gamit, Ghanchi, Vasava and Vania Soni of Gujarat; Kamma, Madiga and Yerukula of Andhra Pradesh.
Populations with low frequency	Keer, Bhiland Gond of Madhya Pradesh; Golla, Mannes, absent in Jatapus, Nooka Dora and Parengi Porojas of Andhra Pradesh.
RH*Cde	
Haplotype frequency	0.63 (0.34-0.96)
Populations with high frequency	STs of India; people of islands; People of Mongoloid affinities of eastern Himalaya, STs of eastern Himalaya, western and north-eastern peninsula; Konda Reddis and Chenchu of Andhra Pradesh.
Populations with low frequency	Indo-European speakers; Brahmins, Koya,Yanadi and Lambadi of Andhra Pradesh.
RH*cde	
Haplotype frequency	0.17 (0.00-0.44)
Populations with high frequency	Indo-European speakers; people of western India.
Populations with low frequency	People of Mongoloid affinities of eastern Himalaya, STs of eastern Himalaya, STs of eastern Himalaya, western and north-eastern peninsula; people of island.
***RH*cDe* (African haplotype)**	
Haplotype frequency	0.056 (0.00-0.41)
Populations with high frequency	People of Mongoloid affinities, Mon-khmer speakers of Austro-Asiatic language family; Konda Reddi of Andhra Pradesh.
Populations with low frequency	STs of Andhra Pradesh.
RH*cDE	
Haplotype frequency	0.09 (0.00-0.03)
Populations with high frequency	Speakers of Tibeto-Burmese and STs of Andhra Pradesh.
Populations with low frequency	Brahmins of Andhra Pradesh.

RH*CDE	
Haplotype frequency	0.01 (0.00-0.13)
Populations with high frequency	Muslims of Uttar Pradesh; STs of Assam and Orissa; Ahirs of Haryana; Muslims of Assam.
Populations with low frequency	Lambadi and Chenchu of Andhra Pradesh.
RH*Cde	
Haplotype frequency	0.02 (0.00-0.17)
RH*CdE	
Haplotype frequency	0.001 (0.00-0.03)
Populations with high frequency	Brahmins and Chenchu of Andhra Pradesh;Irula and Kadar of Tamil Nadu.
Populations with low or nil frequency	Except Chenchu all other STs of Andhra Pradesh.
RH*cdE	
Haplotype frequency	0.004 (0.00-0.09)
Population with high frequency	Brahmins of Andhra Pradesh.
Populations with nil or low frequency	STs of Andhra Pradesh.

3.2.3 MNS System

Forty six antigens were observed in this system. Among them, the following M,N,S,s and U showed higher frequency. Studies with Antisera-S and s, reported that S allele was frequently associated with M than N allele.

MNSs Blood Group	
MN*M (allele frequency)	
Frequency	0.64(0.44-0.89)
Populations with high frequency	People of Himalayan mountain complex, Mongoloid affinity of eastern Himalaya and STs and STs of southern peninsula; Manne,Brahmins, Jalaris, Pradhans and Koyas of Andhra Pradesh; Riang of Tripura.
Populations with low frequency	SCs in Indus-Ganga-Brahmaputra, tribes of western peninsula and speakers of Indo-Europe languages.
MN*N (allele frequency)	
Frequency	0.35
Populations with high frequency	Dhobi of Andhra Pradesh.
Populations with low frequency	Except Dhobi, all populations of India.
Haplotype MNS*MS	
Frequency	0.19
Populations with high frequency	Mikir of Assam; Chenchu of Andhra Pradesh; Sikh of West Bengal.

Populations with low frequency	People of Mongoloid affinities of eastern Himalayan region; speakers of Tibeto-Burmese; Muslims and Christians of Orissa; Khasi of Meghalaya; Onge of Andaman; Chetri, Limboo, Rai, Tamang of Sikkim; Brahmin and Meiti of Manipur.
Haplotype MNS*Ms	
Frequency	0.43
Populations with high frequency	Nicobarese of Andaman; Muslims of Orissa; Bhotia and Lepcha of Sikkim; Kota and Toda, Tamil Nadu; Rajbanshi of West Bengal; Chenchu of Andhra Pradesh;STs of Dadra and Nagar Haveli.
Populations with low frequency	STs of western Peninsula.
Haplotype MNS*NS	
Frequency	0.09
Populations with high frequency	–
Populations with low frequency	People of Mongoloid affinities of eastern Himalayan region; Speakers of Tibeto-Burmese.
Haplotype MNS*Ns	
Frequency	0.26
Population with high frequency	STs of western peninsula, Chenchu of Andhra Pradesh; Kokna and Worli of Dadra and Nagar Haveli; Kota and Kurumba of Tamil Nadu; Siddi of Karnataka.
Populations with low frequency	STs of eastern and southern Peninsula.

3.2.4 P Blood Group System

The P blood group consists of three antigens i.e. P_1, P^{174} and P^K. Depending on the presence or absence of these serological specificities on red blood cells, five phenotypes in P system are reported. They are P_1, P_2,P_1^k, P_2^K and P. P_1 red blood cells (RBC) have P_1 and P antigens, P_2 erythrocytes are deficient of P antigen; and $P_1^k$and P_2^K RBC show P^K activity; P_1 antigen found in P_1^k RBC. Most of the data in Indian populations were available on P_1 antigen.

P Blood Group	
***P*P*$_1$ (allele frequency)**	
Frequency	0.41 (0.40-0.70)
Populations with high frequency	Speakers of Dravidian language; people of south India.
Populations with low frequency	People of Himalayan mountain complex, Mongoloid affinities from Himalayan region and STs of western, north-eastern and southern Peninsula; Caste populations; speakers of Tibeto-Burmese.

3.2.5 Lutheran Blood Group System

This system consists of 25 antigens. Most of the data on Lutheran blood group in Indian population was reported on Lu^a.

Lutheran Blood Group	
***LU*A* (allele frequency)**	
Frequency	0.016
Populations with high frequency	Scheduled tribes of India; speakers of Tibeto-Burmese and Indo-European languages.
Populations with low frequency	Speakers of Dravidian languages.

3.2.6 Kell Blood Group System

In this blood group 25 antigens were reported and these are glycoproteins. Most of the data on Indian populations reported on K antigen.

Kell Blood Group	
***KEL*K* (allele frequency)**	
Frequency	0.038
Populations with high frequency	Ethnic groups of West Bengal; speakers of Indo-European languages; STs of western and southern peninsula.
Populations with low frequency	People of Mongoloid affinity from eastern Himalaya; speakers of Tibeto-Burmese and Dravidian.

3.2.7 Duffy Blood Group System

Six antigens (Fy^a, Fy^b, Fy3, Fy4, Fy5, Fy6) are reported in this system and they are glycoproteins (complex of carbohydrates and proteins). Studies on Indian population reported data on Fy^a.

Duffy Blood Group	
***FY*A* (allele frequency)**	
Frequency	0.053 (0.13-1.00)
Populations with high frequency	People of Mongoloid affinity from eastern Himalaya; STs of southern peninsula; Chenchu and Lambada of Andhra Pradesh.
Populations with low frequency	Speakers of Mon-Khmer and Munda of Austro-Asiatic and Indo-European languages of central, eastern and southern region; Siddi of Karnataka.

3.2.8 Kidd Blood Group System

This blood group contains 3 antigens, Jk^a, Jk^b and Jk3. Studies on India populations were available only on Jk^a antigen.

Kidd Blood Group	
***JK*A* (allele frequency)**	
Frequency	0.052 (0.30-0.81)

Populations with high frequency	Caste populations and speakers of Indo-European language; Rajputs of Himachal Pradesh.
Populations with low frequency	People of Mongoloid affinity of eastern Himalayan; Speakers of Tibeto-Burmese and Dravidian languages; Lambada of Hyderabad.

3.2.9 Diego Blood Group System

In this blood group, 21 antigens were reported. Among them Di^a, Di^b and Wr^a were considered significant.

Diego Blood Group	
***Di*A* (allele frequency)**	
Frequency	0.01 (0.00-0.03)
Populations with high frequency	Populations of Mongoloid affinity of eastern Himalayas; Speakers of Tibeto-Burmese and Indo-European languages.
Populations with low frequency	Sporadic cases in other populations.

3.2.10 ABH Secretor System

ABH secretors are those who secrete antigens (A,B and H) in saliva and mucus. Depending on the blood group type the secretors secrete antigens (A blood group=A and H antigens; B blood group=B and H; O blood group=H only)

ABH Secretor System	
***ABH*Se* (allele frequency)**	
Frequency	0.52 (0.21-0.86)
Populations with high frequency	Populations of Himalayan mountains; SCs of western and north-eastern peninsula; speakers of Tibeto-Burmese and Dravidian languages; Khond of Orissa; people of south India and Islands.
Populations with low frequency	Speakers of Indo-European languages;SCs of south India; Onge of Andaman.

3.3 SERUM PROTEINS

Serum from whole blood is drawn by allowing clotting of the blood, centrifuged and the supernatant is collected. Structural differences in the constant region of the either light or heavy chain of particular immunoglobin is called allotypes, these are inherited and responsible for polymorphism in the serum proteins. These allotypes are encoded by genes. Genes exists as alleles. When alleles exist in combinations, they are called haplotypes. In human, five types of immunoglobulins such as IgG, IgM, IgA, IgE and IgD arefound in serum. Immunoglobulins consist of two light and heavy chains. Both chains composed of variable and constant regions. The constant region for particular class of immunoglobulins remains the same whereas variable region differs from one immunoglobulin to other. The light chain is made up of kappa and lambda chains. Allotypes are observed in heavy chains of IgG1, IgG2, IgG3, IgA2 and IgE and kappa light chains which are encoded by $\gamma 1$, $\gamma 2$, $\gamma 3$, $\alpha 2$, ϵ and κ genes.

The resulting allotypes are labelled as GM, AM, EM and KM. Allotypes in immunoglobulins arises due to mutations in the genes.

Check Your Progress

2) What is allotype and observed in which immunoglobulins?

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3.3.1 GM System (Gamma Marker System)

GM System	
Allotypes and allele or haplotypes	Alleles and haplotypes:*GM*5, GM*1, GM*1,5 and GM*1,2* allotypes were observed.
Frequency of alleles and haplotypes	*GM*5* (0.355), **1* (0.276), *GM*1,5* (0.269) and *GM*1,2* (0.100).
GM*1,5 haplotype	
Populations with high frequency	People of east India and STs, Mongoloid affinity of eastern Himalyan region and speakers of Tibeto-Burmese, Munda section of Austro-Asiatic languages and north and central region of Dravidian speakers.
GM*5 alleles	
Populations with high frequency	Speakers of Indo-European and Dravidian languages.
GM*1*, ***GM*1,5 and ***GM*1,2 alleles and haplotypes***	
Populations with high frequency	People of Himalayan mountain complex.

3.3.2 Kappa Marker (KM) System

KM System	
Allotypes and Alleles	KM1, KM2, KM3; alleles *KM*1, KM*1,2, KM*1,3*
Frequency of allele	*KM*1*0.11(0.021-0.032)
KM*1	
Populations with high frequency	People of Himalayan mountain complex and Mongoloid affinity of eastern Himalaya, STsof western and north-eastern peninsula; speakers of Tibeto-Burmese language.
Populations with low frequency	Speakers of Indo-European and Dravidian languages; STs of southern peninsula.

3.3.3 Haptoglobin(HP) System

Haptoglobin System	
Protein information	Haptoglobin is a glycoprotein and consists of 2 α and 2 β chains. Polymorphisms in α and β reported but most of the data was reported in α chain only.
Phenotypes and alleles	Chain Phenotypes alleles **α** HP1, HP2-1, HP2 *HP*1, HP*2*
HP*1	
Frequency	0.16 (0.00-0.40)
Populations with high frequency	People of Himalayan mountain complex; STs and people of Mongoloid affinity of eastern Himalaya; Speakers of Mon-Khmer of Austro-Asiatic, Tibeto-Burmese and Indo-European languages: Chenchu, Vysyas, Kammas and Madigas of Andhra Pradesh; Ladakhi of Jammu and Kashmir; Siddi and Jenu-Kerumba of Karnataka; Ezhava of Kerala.
Populations with low frequency	STs of Brahmaputra plains, western and north eastern peninsular plateau, South India; Speakers of Munda of Austro-Asiatic and Dravidian languages; Manne and Yerukula of Andhra Pradesh; Santal of West Bengal.

3.3.4 Transferrin(TF) System

Transferrin System	
Protein information	Transferrin is an iron binding glycoprotein and transport iron in the blood.
Phenotypes	TFC (C1,C2,C3,C4,C5,C6,C7,C8,C9,C10,C11,C12, C13), TFD (D,Chi), TFB(B).
Frequency	*TF*C* (0.991), *TF*D* (0.008) and *TF*B* (0.001).
TF*C1	
Populations with high frequency	Western India, Brahmins, Konda Kamaras,Koyas, Lambada and Vysyas of Andhra Pradesh.
Populations with low frequency	People of Mongoloid affinity of Assam and Manipur.
TF*C2	
Populations reported	Vysyas, Koyas and Lambadi of Andhra Pradesh.
TF*C3 (European marker)	
Populations with high frequency	North Indian population;Konda Kamaras,Koyas, and Lambada of Andhra Pradesh.
TF*D	
Populations with high frequency	SC and ST population, tropical savannah type climate region; speakers of Munda of Autstro-Asiatic and Dravidian languages, Madiga, Maria Gond, Lambada and Koyas of Andhra Pradesh.
TF*Chi	
Populations reported	Oraons of Bihar.

TF*B	
Populations reported	Jalari and Madiga of Andhra Pradesh.

3.3.5 Group Specific Component System (GC)

Group Specific Component System(GC)	
Protein information	α 2 globulin
Phenotypes	GC 1S, GC 1S1F, GC 1F,GC 2-1S, GC 2-1F and GC 2; alleles *GC*1(GC*1F and GC*1S) and GC*2*
GC*1	
Frequency	0.74 (0.59-0.91)
Populations with high frequency	Munda speakers of Austro-Asiatic languages; Mala, Madiga, Rajgonds, Pradhans and Muslims of Andhra Pradesh; Karabi of Assam, Brahmin, Chetri and Pradhan of Sikkim; Langia-Saora of Orissa and Brahmin of Karnataka.
Populations with low frequency	People of Himalayan region; ST of Indus-Ganga-Brahmaputra; speakers of Indo-European languages.
GC*1S	
Frequency	0.49
Populations with high frequency	People of Mongoloid affinity of western Himalaya and Onge of Andaman.
GC*1F	
Frequency	0.25
Populations with high frequency	People of Mongoloid affinity of eastern Himalaya; Siddi; people of northern and southern region of India.

3.4 BIOCHEMICAL OR RED CELL ENZYME/ PROTEIN MARKERS

3.4.1 Adenosine Deaminase (ADA) System

Adenosine Deaminase System	
Enzyme information	Enzyme commission No**.** 3.5.4.4. It is a aminohydrolase. The role of this enzyme is to catalyze the deamination of adenosine to inosine.
Phenotypes and alleles	ADA1, ADA2, ADA2-1 and alleles: *ADA*1*, *ADA*2*
ADA*1	
Frequency	0.88 (0.50-0.98)
Populations with high frequency	People of western, north-eastern and southern region; Hindus, Jalari, Lambada and Savara of Andhra Pradesh.

Populations with low frequency	People of Indus-Ganga-Brahmaputraplain region; STs and people of Mongoloid affinity of eastern Himalayan region. speakers of Indo-European languages; speakers of Tibeto-Burmese.

3.4.2 Adenylate Kinase (AK) System

Adenylate Kinase (AK) System	
Enzyme information	Enzyme commission No. 2.7.4.3. The role of this enzyme is to catalyze the interconversion of adenine nucleotides such as Adenosine triphosphate, Adenosine monophosphate and Adenosine diphosphate.
Phenotypes and alleles	Adenylate Kinase1, Adenylate Kinase2, Adenylate Kinase3; alleles: *AK*1*, *AK*2*and *AK*3*
AK*1	
Frequency	0.92 (0.79-1.00)
Populations with high frequency	People of Himalayan mountain complex; STs of Mongoloid affinity of eastern Himalyas; speakers of Tibeto-Burmese; Jalari, Hindu, Sugali and Kolam of Andhra Pradesh; Ezava and Pulayan of Kerala
Populations with low frequency	STs of western, northern and southern peninsula; speakers of Indo-European, Munda group of Austro-Asiatic and Dravidian languages.

3.4.3 Acid Phosphatase (ACP) System

Acid Phosphatase (ACP) System	
Enzyme information	Enzyme commission No 3.1.3.2**.** It is a phosphohydrolase. The role of this enzyme is to catalyze the hydrolysis of phosphatase in acidic environment.
Phenotypes and alleles	ACP1A, ACP1BA, ACP1CA and ACP1CB; alleles: *ACP1*A*, *ACP1*B* and *ACP1*C*
Frequency	*ACP1*A*(0.10-0.36), *ACP1*B* (0.64-0.86) and *ACP1*C* (0.00-0.0035)
ACP1*A	
Populations with high frequency	SCs of Indus-Ganga-Brahmaputra plain region; STs of southern and western peninsular plateau; speakers of Indo-European; Muslim, Chenchu, Jalari, Vysyas and Madiga of Andhra Pradesh.
Populations with low frequency	SCs of southern peninsular plateau; STs of north-eastern peninsular plateau and people of Mongoloid affinity of eastern Himalaya.
ACP1*C	
Population reported	People of Himalaya region.

3.4.4 Phosphoglucomutase (PGM) System

Phosphoglucomutase (PGM) System	
Enzyme information	Enzyme commission No 2.7.5.1**.**It catalyze the interconversion of glucose-6-phosphate and glucose-1-phosphate.
Phenotypes and alleles	PGM1 1-1, PGM1 2-2, PGM1 2-1; alleles: *PGM1*1* and *PGM1*2*
PGM1*1	
Frequency	0.70 (0.44-0.95)
Populations with high frequency	SCs of southern peninsular region; STs of western and north-eastern peninsular plateau; speakers of Dravidian, Indo-European and Tibeto-Burmese; STs of West Bengal, Karnataka and Andhra Pradesh; Oriya and SaoraLangia of Orissa; Lingayat and Vokkalinga of Karnataka; Bagdi, Kaibarta and Kaora of West Bengal, Bhil and Meena of Rajasthan; Nadar, Reddiar and Thevar of Tamil Nadu.
Populations with low frequency	Southern peninsular region; STs of Indus-Ganga-Brahmaputra plain region, southern peninsular region and eastern Himalaya; speakers of Munda group of Austro-Asiatic languages; STs of north India; Yerukula of Andhra Pradesh.

3.4.5 Phosphogluconate Dehydrogenase (6-PGD) System

Phosphogluconate Dehydrogenase (6-PGD) System	
Enzyme information	Enzyme commission No 1.1.1.43**.** It catalyse the oxidative decarboxylation of 6-phosphogluconate to ribulose-5-phosphate.
Phenotypes and alleles	PGDA, PGDAC, PGDC; alleles: *PGD*A* and *PGD*C*
PGD*A	
Frequency	0.95 (0.75-1.00)
Populations with high frequency	SCs of India; STs of southern and north-east peninsular region; people of Mongoloid affinity from eastern Himalays; Brahmins, Konda Reddy, Jatapu, Pradhan, Savara and Muslims of Andhra Pradesh.
Populations with low frequency	People of Himalayan mountain complex; speakers of Bhotia and Himalya group of Tibeto-Burmese and Pahari of Indo-European languages.

3.4.6 Esterase D (ESD) System

Esterase D (ESD) System	
Enzyme information	Enzyme commission No 3.1.1.1**.** It is serine hydrolaze and involved in detoxification of formaldehyde.
Phenotypes and alleles	ESD1-1, ESD 2-2, ESD2-1; alleles: *ESD*1* and *ESD*2*

*ESD*1*	
Frequency	0.72 (0.41-0.97)
Populations with high frequency	People of Himalayan mountain complex; SCs of Indus-Ganga-Brahmputra; STs of western peninsular plateau; speakers of Indo-European language.
Populations with low frequency	People of southern and north-eastern peninsular plateau; STs of Mongoloid affinity from eastern Himalayas; speakers of Munda group of Austro-Asiatic, Bodo and Kuki of Tibeto-Burmese and north andcentral groups of Dravidian language families.

3.4.7 Glucose-6-Phosphate Dehydrogenase (G-6-PD) System

Glucose-6-Phosphate Dehydrogenase (G-6-PD) System	
Enzyme information	Enzyme commission No 1.1.1.49. It catalyzes the reduction-oxidation of glucose-6-phosphate.
Phenotypes and alleles	G6PD A and G6PD B allele: G6PD*B,G6PD* Andhra Pradesh, G6PD*Cutch, G6PD* Jammu, G6PD* Kalayan, G6PD*Kerala, G6PD*Porbandar and G6PD*West Bengal.
G6PDeficiency	
Frequency	0.045 (0.00-0.27)
Populations with high frequency	People of Himalayan region; STs of Mongoloid affinity from eastern Himalayas; north and western region; people of Andhra Pradesh and Tamil Nadu; speakers of Mon-khmer, Bodo group, north-east frontier group and Pahari group of Himalayas; Angami Naga; Adi, Apatani and Nishi of Arunachal Pradesh.
Populations with low frequency	STs of southern peninsularplateau; People of southern and north-eastern peninsular plateau; speakers of Dravidian language; Brahmins, Golla, Mala, Manne, Madiga, Koyas and Pradhan of Andhra Pradesh; Rajput of Himachal Pradesh; Gond and Oraon of Madhya Pradesh; Santal of West Bengal.

3.4.8 Hemoglobin (HB)

Hemoglobin (HB)	
Protein information	It is a tetramer protein and contains two alpha and two beta chains. HB transport oxygen, nitric oxide and carbon dioxide in the body.
Phenotypes	HBS, HBD and HBE; alleles: *HB*S,HB*E* and *HB*D*
HB*S	
Frequency	0.031 (0.00-0.41)
Populations with high frequency	STs and SCs; semi-arid steppe and tropical savannah type climate; south, central, west and north.
Populations with low frequency	East India; Islands.

*HB*E*	
Frequency	0.023 (0.00-0.64)
Populations with high frequency	People of Mongoloid affinity of eastern Himalaya; speakers of Mon-khmer group of Austro-Asiatic and Tibeto-Burmese languages; people of Indus-Ganga-Brahmaputra plain region
*GC*1F*	
Frequency	0.25
Populations with high frequency	People of Mongoloid affinity of eastern Himalayas; Siddi; people of northern and southern region of India.

Check Your Progress

3) What level of protein structure found in haemoglobin, what are the chains it contains and functions of haemoglobin?

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Review of genetic, serological and biochemical markers suggested that a greater genetic variations were observed not only between but also within Indian populations; the amount of genetic diversity in Indian castes was comparable to the races of the world; genetic homogeneity was more in geographically closer populations than speakers of language families or caste/social hierarchy; tribal populations were genetically and morphologically heterogeneous than non-tribal populations and within tribal populations, tribes from south were different from central and north-eastern regions; the genetic diversity among castes was 1-3% whereas between castes and tribes it was 5%; in traits such as ABO, Sickle cell, HBE and G6PD presence of geographical gradation (clines) suggested the role of selection; genetic drift, founder effect, gene flow, fission and fusion were found to be played a greater role in causing micro evolutionary differences in the populations of India and after Africa, greater genetic diversity was observed in Indian populations (*Reddy,* 2008).

3.5 MOLECULAR/DNA MARKERS

Molecular/DNA markers can be categorized into mitochondrial DNA and chromosomal/genomic DNA markers (Y chromosome/autosomal).

3.5.1 Mitochondrial DNA(mtDNA)

It is a circular shaped, double stranded molecule and inherited from the mother to the children. Due to unique characteristics of mtDNA such as high mutation rate, high copy number and lack of recombination, it is used for evolutionary and polymorphism studies. Maternal history of population (migration of women gene pool) can be known using mtDNA. Two approaches used in mtDNA studies are lineage and population based. The former unearths the history of mtDNA lineages or haplogroups while the latter employing population as unit

and population genetic methods reflect prehistory of migrations/geographical areas/populations.

3.5.1.1 Markers Used and Observations of mtDNA Studies

Markers Used in Mitochondrial DNA Studies
1) Hypervariable Regions I & II: These are sites 400bp each present in the 1.1Kb non-coding control region which has regulatory functions (*Sharma et al*. 2005)
2) Intergenic COII/ tRNA(Lys) 9 bp deletion Polymorphism (Haplogroup B): It is characterized by loss of one of two repeated copies of the sequence (CCCCCTCTA) from a non-coding region located between cytochrome oxidase II (COII) and $tRNA^{Lys}$ gene (*Ren et al*.2016).
3) Haplogroups: These are groups of related sequences sharing mutations and ancestry, geographically specific and useful to estimate the percent of admixture in populations belonging to diverse geographical regions and occupants of known routes of migration (*Pakendorf and Stoneking,* 2005).
4) Haplotype: A haplotype is a set of DNA variations, or polymorphisms, that tend to be inherited together (https://www.genome.gov/genetics-glossary/haplotype).
Observations
• Most of the Indian populations belong to haplogroup M. This haplogroup entered in to Asia from east Africa through southern route approximately 60,000 years before.
• Haplogroup U was contributed by western Eurasians to Indian population by multiple migrations.U2i, a sub lineage of U was observed with higher frequency among tribes(>77%). Prehistory of Indian populations 40,000-60,000 was suggested based on macro-haplogroups M and N.
• There is no consensus among authors on the origin and sharing of west Eurasian haplotypes among tribes and castes.
• Correlation between mtDNA distances of castes with social ranking was observed due to the lower caste women marrying higher caste men.
• Mundari group of Austro-Asiatic speakers was established as original inhabitants of India and their ancestors entered into India through western corridor; tribes of Mon-Khmer as having Asian and Mundari group of Austro-Asiatic speakers as non-Asian origin and affinity of north-western Indians to west Eurasians was reported.
• mtDNA diversity was found to be higher than Europeans and East Asians and occupied between continental and African populations. Indo-European speakers shared their mtDNA to the north Indian population.
• Frequency of 9bp deletion was found to be 0.8% in northeast;1.5% in south India and 45.8% in Nicobarese.
• The genetic link between Indians and Indo-Aryans was dated to approximately 50,000 years. Highest frequency of macrohaplogroup M and hypervariable sequence 1 of mtDNA was observed in tribes speaking Austro-Asiatic language.
• Indian populations received restricted gene flow from outside and castes & tribes are descendants of Pleistocene southern and west Asians.

- Indians share 20-30% haplotypes of Eurasians which contributed to rank and sex specific differences in genetic affinity of castes to Europeans and Asians.
- Upper castes than lower castes were found to be genetically related to Europeans than to Asians. mtDNA diversity was greater in north than south Indian tribes and Indian mtDNA gene pool was homogenous to east against west Eurasians.
- Populations of Indus valley showed mtDNA lineage of western Eurasian while inhabitants of Indo-Gangetic region harboured mixed mtDNA lineages of western and eastern Eurasians and south Asians.
- N macrohaplogroup along with European haplogroups such as H,V,K,U5,J and T entered into Indian populations in less than 11.5 thousand years ago.
- It was estimated that three mtDNA lineages at different time periods entered into the Indian subcontinent.
- Genetic link between Khasis and north eastern populations of India was also observed.
- Shia and Sunni groups of Muslims were descents of Indian maternal lineage only and homogenous in terms of mtDNA haplogroups as wellas with other populations of India but differed with Muslims of other regions.
- Maternal lineages of M31,M32 in Onge and greater Andamanese were considered as isolated lineage and not affined with any other group.
- Unique mtDNA haplogroups linked 9bp deletion were observed in Nicobarese and were found to have affinity with Mon-Khmer speaking Cambodian populations.
- B5a and R12 haplogroups of R lineages were found in Shompen tribe of Nicobar Islands. It was observed that Negrito element in people of Andamanese was due to convergence than genetic affinity with Africans.
- mtDNA studies on Indian populations showed single matrilineal origin of castes and tribes and resulted from admixture of populations with other populations and operation of evolutionary forces such as genetic drift.

3.5.2 Y Chromosome

Y chromosome studies in human population are done to infer origin, history and migration routes of male lineage and to reconstruct evolutionary history of populations. Y chromosome is transmitted from father to son. Y chromosome has certain unique features such as lack of homologous partner for cross over and recombination of genes, specialized for spermatogenesis and higher percent of repetitive sequences in the DNA. The euchromatic region(actively involved in the transcription of DNA to mRNA) of human Y chromosome is about 23 Megabases and harbour 78 protein coding genes.

3.5.2.1 Markers Used and Observations of Y Chromosome Studies

Marker Used in Y Chromosome Studies
1) Single nucleotide polymorphism: It is also called SNP, is a variation at a single position of DNA sequence among individuals (https://www.nature.com/scitable/definition/snp-295/)

2) Microsatellites or Short tandem repeats (STR): It contains repeats of 1-6 base Pairs (Kayser et al. 2004). They are used either to discriminate or infer lineage relationships. In evolutionary studies upto 16 Y chromosome satellites were used.
3) Haplotype: A haplotype is a set of DNA variations, or polymorphisms, that tend to beinherited together (https://www.genome.gov/genetics-glossary/haplotype).
4) Haplogroup: Please refer mtDNA marker list.

Observations

- Patterns of variation in terms of haplogroups were found to be distinct between tribes and castes.
- There is no consensus among authors on the genetic flow of paternal lineage among Indian populations.
- No correlation of caste rank with genetic distances based on Y chromosome data was found but genetic affinity of eastern Indo-European speakers with higher caste was observed. Higher frequency of haplogroups such as HG-3 and HG-9 among north Indian populations; haplogroup R1a among Punjabi and Chenchu of Andhra Pradesh and haplogroup K in Austro-Asiatic speakers, was noted.
- No sex specific variations based on Y chromosome data among patri or matrilocal groups was noticed.
- Authors of Y chromosome studies reported no recent admixture in caste populations; placed north Indians as intermediate between west and central Asian populations; observed genetic affinity of caste populations to central Asians and castes of south India to east Europeans; recent admixture of Indo-Europeans speakers in Indian population; genetic linkage of northeast Indians and Chinese and migration of Austro-Asiatic speakers of India to south east Asia through northeast India corridor.
- Lower genetic variation among Andamanese and their genetic affinity with Asians was reported.
- Among Shompen tribes, existence of single lineage (O-M95) and their genetic affinity with Nicobarese, Austro-Asiatic speakers of India and south-east Asia was found.
- Genetic link between Indians and Australians was dated back to Holocene.
- Muslims were closely related to non-Muslim neighbours than the followers of their religion from other regions.

3.5.3 Autosomes

mtDNA or Y chromosome studies show only uniparental and gender specific origin, history and migration pathways, represent lower percent of individual ancestry and influenced by genetic drift which cause fluctuations in the frequencies of mutations in populations over generations. Effect of drift on autosomes was found to be small extant and its data is genomic representatives of both parents. Recombination occurs in alleles and assorted independent of each other and passed on to the next generation. Autosomes provide information on history and prehistoric admixture among the populations. Compared to mtDNA and Y chromosome, larger number of genes are found on the autosomes.

3.5.3.1 Markers Used and Observations of Autosomal Studies

Marker Used in Autosomal Studies
1) Short tandem repeats (STR): These are located around the centromere of autosomes (22 pairs and each individual inherit one copy each from mother and father) for responsible for somatic characteristics of individuals. They are 2-6 base pairs in length which are repeated over and over in a defined location of autosomes of chromosomal DNA. 2) Insertion/deletion polymorphism: They are also called indels which involve deletion or insertion of specific nucleotide sequence. A total of 3 million indels are observed in human genome. 3) Single nucleotide polymorphism: Please refer Y chromosome marker list. 4) Haplotype: Please refer mtDNA marker list. 5) Haplogroup: Please refer mtDNA marker list.
Observations
• Autosomal markers were used for studying genetic affinities between populations of India. • No consistent trend in studies on autosomal markers linking genetic distance to geographic, linguistic, ethnicity and social ranking was observed in Indian populations. • Autosomal haplogroups MX1 and Ch21 in Koya, Chenchu of Andhra Pradesh along with other Indian caste populations showed clear distinction from European and east Asian populations. • Populations from south India showed lower heterozygosity when compared to north Indian populations. • Genetic affinity was greater among geographically closer populations. • Indian genome variation studies on 405 SNPs from chromosome 22 in 1871 individuals belonging to 55 populations placed Indian populations as genetically intermediate between west and East Asian populations.
• HUGO Pan Asian SNP consortium study reported genetic affinity of India populations with west Asians. • High ranking caste populations of north India were found to be genetically closer to central Asian populations. • It was observed that ancient human populations migrated to India through southern and northern exits and formed Ancestral North Indian (ANI) and Ancestral South Indian (ASI) populations. All extant Indian populations are descendants of admixed two ancestral populations. Higher contribution of ANI in north Indian and ASI in south India was observed. • In Austro-Asiatic speakers, ancestral Austro-Asiatic (AAA) of central and east India and ancestral Tibeto-Burman(ATB) among Tibeto-Burman speakers of north east were identified. ANI and ATB were proposed to be co-ancestral to central south Asians and south East Asians. Both ASI and AAA populations were considered as early settlers in India due to lack of genetic affinity of them with neighbouring populations presumably entered into India through southern exit.

- During 1900-4200 years before past admixture occurred between populations in India and after that endogamy became standard social practice.

3.6 SUMMARY

In this unit polymorphisms reported in Indian populations on blood groups, serum proteins, red cell enzymes and protein and molecular markers of mtDNA, Y chromosome and autosomes were reviewed. Genetic/serological/biochemical markers showed greater diversity in caste populations, genetic homogeneity among geographically closer populations and operation of selection, genetic drift, gene flow, fusion and fission played a greater role in causing variations in Indian populations. mtDNA studies suggested single matrilineages of tribes and castes and variations due to admixture with other populations and operation of genetic drift. Y chromosome haplogroup frequencies were distinct between tribes and castes and admixture of Indian populations with east and west Eurasians, south and central Asians and migration of Austro-Asiatic speakers to southeast Asia through northeast Indian corridor. Autosomal markers were employed to infer genetic affinities between populations.

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3.8 ANSWERS TO CHECK YOUR PROGRESS

1) Variations in humans is caused by waves of migrations and admixture with indigenous people, presence of racial elements, adaptations to the environment, isolation, mating patterns and operation of evolutionary forces such as mutations, gene flow, genetic drift, recombination and natural selection. Study of variations are useful to investigate evolutionary relationships, evolutionary history, gender specific (mitochondrial DNA and Y chromosomes) migrations, appreciate the extent of variation in humans, enhance our knowledge on evolutionary forces, susceptibility (BRCA1 and BRCA2 genes increase the risk of breast or ovary cancer) or protection (mutations in CCR5 gene protect against HIV) to diseases and drug responses (P450CYP2CP gene variants and Warfarin drug dose requirements).

2) Structural differences in the constant region of the either light or heavy chain ofparticular immunoglobin is called allotypes. Allotypes are observed in heavy chains of IgG1, IgG2, IgG3, IgA2 and IgE and kappa light chains.

3) In haemoglobin, quaternary level of protein structure is observed. It contains 2 alpha and 2 beta chains. Hemoglobin transport oxygen, nitric oxide and carbon dioxide in the body.

UNIT 4 ROLE OF BIO-CULTURAL FACTORS*

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4.4 Summary

4.5 References

4.6 Answers to Check Your Progress

Learning Objectives

After reading this Unit, you will be able to:

- Appreciate the role of bio-cultural factors influencing the diseases and nutritional status;
- Discuss the details of evolution of human diet; and
- Elucidate biological perspectives of aging process among different populations.

4.0 INTRODUCTION

Since the mid-twentieth century, the bio-cultural approach has served as an integrative intellectual approach within anthropology, particularly within the sub-disciplines of biological, medical, and socio-cultural anthropology. It has provided an avenue for synthetic research that unites and cross cuts these diverse arenas, helping to prevent fragmentation and schism in the face of increasing specialization. Further, it enables anthropologists to achieve the core anthropological objectives of explaining human behaviour across time and space, comprehending cultural similarity, difference and complexity across space and time, and applying this knowledge to the solution of human problems.

McElroy (1990) defined humans as the biological, social and cultural beings in response to the environment. Though bio-cultural aspect has been intrinsic part of physical/biological anthropology but it gained currency when the Human

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adaptability project of International Biological Programme was initiated in 1960's (*Weiner,* 1964) and socio-cultural as well as biotic environment was made as a part of research design to investigate human variation. Demographic, genetic, anthropometric, blood protein and blood pressure variables were used to study the relation between biology and environmental factors such as cultural, behavioural, psychological, socio-economic, historical and ecological.

Bio-cultural studies employ variables such as biological, cultural (behavioural and psychological) and environmental (biotic (living) and abiotic (non-living)). Bio-cultural studies, explore the relationship between these variablesat the population level particularly those contributing to the interaction of biology and culture and also attempt to understand human variation. Bio-cultural approach bridges gap between sub disciplines of biological/physical and social/cultural anthropology and promote integrative thinking in addressing the problems faced by the society. Biological anthropologists are more comfortable in observing (morphological) and measuring (biochemical and physiological) biological variables. It is interesting to note thatthese variables are influenced by demographic variables (age, gender, education, income, marital status, religion and occupation), socio-economic conditions, behavioural (smoking and alcoholism) and cultural factors (diet and physical activity). Biological anthropological studies should collaborate with the social anthropological studies to carryout bio-cultural studies and also to know which socio-cultural variables influence human biology (*Khongsdier,* 2007).

4.1 BIO-CULTURAL FACTORS INFLUENCING THE DISEASES AND NUTRITIONAL STATUS

The influence of bio-cultural factors on diseases and nutritional status is discussed below:

4.1.1 Diseases

Disease is a pathological state of the organism, without regard to cultural or psychological acceptance. Disease was considered as western concept whereas illness is culturally defined and diagnosed using traditional knowledge. Culture not only help to understand the health condition but also assist with available resources to cope, protect and prevent by adjusting physical and social environments. Investigation of people of different culture may inform native system of disease identification, nomenclature and other related issues. It was observed that there are universal perceptions and society specific perceptions with reference to health and disease. At each step of suffering of causation, experience and expression of suffering, the contribution of culture is recognized.

Though the concept of health and illness are biological but they are also related with socio-cultural contexts. As a result, in health studies, beliefs and pattern of disease connected with health and disease should be included. Certain symptoms were found to be unique to specific cultural groups known as culture bound syndromes. Examples of the syndromes are 1. Koro, a condition in which people believe that sexual organs are shrinking which is observed among people of Southern China and South-East Asia; 2. Latah syndrome, itinitially begins as an exaggerated amazing response to a surprising event and later result into

a lifelong condition. This syndrome is noticed among the people of Malaysia and Indonesia 3. Bebinan, this syndrome was found among the people of Bali. Sufferers of this syndrome suddenly start crying, run away, collapse after exhaustion and later forget all the happenings and 4. Tabacazo syndrome was reported among the people of Chile. The cardinal symptoms of this syndrome are loss of consciousness with aggression, agitation and despair.

Comparative studies on mental health inform different view points on mental disorders in different cultures. For example, in case of suicide, culture act as either facilitator or protective factor against suicides. Goldstan et al (2008) reported that among adolescent immigrants or adolescents belonging to immigrants to United States, pressures and stresses to assimilate the adapted country culture while retaining their cultural identity act as facilitator of suicides whereas access to community and extended family support, traditional activities and spirituality serve as a protection against suicides among adolescent African/Indian Americans. Some genetic or acquired diseases are found in all cultures (*Hassan,* 2012) and some disease like non-communicable diseases (NCDs) such as cardiovascular diseases, diabetes and chronic respiratory disease are caused by exposure to risk factors like tobacco and alcohol consumption, physical inactivity, overweight, high blood pressure and high blood cholesterol which are socially patterned. High burden of these risk factors was shown to be shifting from high to lower income countries due to higher consumption of tobacco, alcohol, less healthy diet such as less fish, vegetables, fibres and more meat in low income people and physical inactivity in higher income folk (*Stringhini* and, *Bovet,* 2017). To better understand the role of bio-cultural factors in diseases, the following examples using Kuru, Sickle cell anaemia, Lactate intolerance and Pibloktoq diseases, are described.

4.1.1.1 Kuru

Kuru, a prion disease (accumulation of 'prion' proteins in the brain).This disease was reported in Fore tribe belonging to Papua New Guinea. It caused 1000 deaths during the period of 1957-61. The disease transmission from one person to other was reported due to the practice of endocannibalism, a ritual of eating the dead bodies of relatives contracted to Kuru disease. This disease was characterized by degeneration and subsequent loss of neurons associated with vacuolization, astrocytic (star shaped cells of the brain) hypertrophy (enlargement of cells) and proliferation (increase in cell number), spongiform (sponge like) change, and presence of amyloid protein clumps. After doing away the ritual of endocannibalism, the decline in the occurrence of Kuru was reported (*Alpers,* 2008).

4.1.1.2 Sickle Cell Trait

The name of this disease is derived due to sickle shape of haemoglobin(HGB), the carrier protein of oxygen to the cells of body tissues in the body. This is due to mutation in the gene for beta globin of HBG as a result of which amino acid 'Valine' is replaced with 'glutamic acid' in sixth position of Beta globin gene. Carriers of sickle trait (SCT) inherit the abnormal sickle cell gene from one of the parents and normal beta globin gene from another parent. Normally red blood cells (RBC) are discoid in shape and move easily in the blood vessel. When they assume sickle shape, they block the blood vessel and increase the

risk of stroke, infections, episodes of pain (pain crises) and eye abnormalities. The symptoms of carriers of SCT are yellow skin, swelling of limbs and whites of eyes.Frequency of SCT is higher in Western Africa. It was noted that frequency of SCT was lower in hunter and gathering people than those dependent on agriculture. When forests were cleared for the development of agriculture which provided breeding ground for the transmission of Plasmodium falciparum, a protozoan parasite responsible for the malaria. People with SCT are heterozygotes for Sickle Cell gene and are resistance to the infection of malaria than normal people or homozygotes for the sickle cell disease (*Khongsdier,* 2007).

4.1.1.4 Lactase Intolerance

In population in which dairy products are major dietary components continue to synthesize the enzyme lactase-phlorizin hydrolase beyond childhood. This enzyme is responsible for the digestion of lactose (carbohydrate) present in the milk. This phenomenon is called lactase persistence and inherited as autosomal dominant trait. Approximately 35% of people was shown to possess lactose persistence haplotype across the world with the highest frequency in Americans of European origin and European residents and lowest frequency among South-East Asians and Sub-Saharan Africans (*Hassan et al.* 2016). Those who fail to produce adequate levels of the enzyme lactase phlorizin suffer from abdominal pain, diarrhoea, flatus, borborygmi and distension. This is known as lactase intolerance. The frequency of lactase intolerance was reported to be approximately 65% in World populations being most common among African Americans, Asians and Hispanics/Latinos and the lowest frequency has been noted among people of European origin (*Malik* and *Panuganti*, 2020).

4.1.1.4 Pibloktoqor Arctic Hysteria

This aberrant behaviour is noticed among Eskimos. This condition is characterized by four phases. First phase or prodrome lasts hours to days containing irritation and social withdrawal; second phase stay 30 minutes and involves symptoms like sudden onset of extreme and wild excitement, running in the streets, shouting, tearing off clothes and throwing available objects;third phase may stay for 12 hours and consists of fits, stupor, calmness or comma; and final phase is characterized by complete amnesia (*Fulk*, 2012). This condition is caused by hypervitaminosis A due to higher consumption of liver and fats of arctic and marine animals, which are rich sources of Vitamin A in traditional diet of Eskimo and storage of Vitamin A in poisonous quantities. This is a good example of interaction of biology and cultural factors (*Khongsdier*, 2007; *Landy*, 1985).

Check Your Progress

1) What is Bio-Cultural Anthropology?

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2) What is Pibloktoq?

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4.1.2 Nutrition

Food consumption ensures availability of essential nutrients to the body and its expenditure provides energy to perform physical, mental and reproductive roles and maintenance of health. Malnutrition may cause ill health and diseases. Culture facilitates the availability and consumption of food. Nutritional anthropology explores the role of biological and social forces in the usage of food and also investigate the nutritional status of populations. Jerome et al. (1980) explained ecological model for food and nutrition and opined that environmental (climate, soil properties, fauna and flora and water resources) and social sectors/factors (social organizations, technology (tools and techniques), culture (religious beliefs on food, ideas on food and health, food preferences/restrictions and, use of food in social interactions) influence the nutrition of people. Environment sector/factor provide conditions for production and procurement of food while the social sectors facilitate the production and distribution in social groups (*Pelto et al.*2000).

The bio-cultural approach emphasizes the investigation of interaction of both sectors/factors. Interaction of biology and culture takes place through genetic, physiological and socio-cultural adaptations. Genetic adaptations are passing of genes from one generation to other through hereditary that improve the survival of organisms with advantageous genes. This phenomenon is explained by the studies of differences in the ability of older children and adult populations in digesting milk without gastric disturbances. During the time of chronic caloric deficits, one form of physiological adaptation is reducing the basal metabolic rate. Growth stunting observed in early childhood is a physiological adaptation to inadequate food. Socio-cultural adaptations consist of behavioural and technological innovations. Development of manioc processing to better utilize bitter manioc and maize processing techniques helpful in preventing pellagra can improve the ability of people to utilize food resources are the examples of cultural adaptations (*Pelto et al*.2000)

Agriculture though improves the yields of energy but this innovation involves other costs also. Investigation of food patterns of long time periods suggested that they were positive cultural adaptations which maximized nutritional prosperity. The negative aspects of biological and cultural adaptations came to the notice were denial of food to certain sections of society such as women and children; maintenance of discriminatory food patterns by enforcing of non-verbal and informal customs by the followers of certain practices; and imposing of food prohibitions through traditional beliefs and food restrictions. Practice of food prohibitions was reported as a cause of vitamin A deficiency/xerophthalmia in the children of Malaysia (*McKay*,1971; *Pelto et al*.2000). The role of bio-cultural factors in nutrition are explained by nutritional Rickets, low prevalence of ulcer among Garo and malnutrition.

4.1.2.1 Nutritional Rickets

This occurs due to softening and weakening of bone in children (https://www.nhs.uk/conditions/rickets-and-osteomalacia/) and caused by low vitamin D/sun exposure/calcium intake (WHO, 2019). Nutritional Rickets was recognized as a significant problem among Asian immigrants in United Kingdom (*Black*, 1989; *Hassan*, 2012). Factors like less exposure to sunlight due to less coverage of limbs, vegetarian diet, poor intake of vitamin D, maternal vitamin D deficiency and feeding of infants with vitamin D poor Cow's milk were found to be responsible for Rickets among them (*Black*, 1989). This is good example of interplay of cultural factors and physical disease.

4.1.2.2 Low Prevalence of Ulcer Among Garo of North-East India

Garo tribal people consume rice with curries of vegetables, pulses, meat and egg. They boil most of their food item and rarely consume fired or spicy food. They drink alkali water extracted from plant ash which neutralize the acid production in the stomach. The low prevalence of ulcers among Garo was considered to be due to their dietary habits and type of cooking which is a good example of culture acting as protective factor against ulcer disease (*Hassan*, 2008).

4.1.2.3 Malnutrition

This condition is due to deficiencies, excesses or imbalances in a person's intake of energy or nutrients (WHO, 2016). It causes decreased working capacity leading to poor productivity, increase poverty, low standards of living and illness and disease. Malnutrition is caused by increased energy expenditure, reduced dietary intake, reduced absorption of macro and micronutrients and increased losses or changed requirements (*Saunders* and *Smith*, 2010) which affects their body fitness and results in maladaptation to the working demands in less mechanized and labour intensive tasks which influences other components of wellbeing (*Khongsdier*, 2007).

Check Your Progress

3) What is Malnutrition?

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4.2 EVOLUTION OF DIET

Evolution of human diet consists of four stages. 1. Miocene to early Pleistocene 2. Paleolithic 3. Neolithic and 4. Industrial revolution (*Jew et al*. 2009). In Miocene to early Pleistocene human diets were of foliage, nuts, seeds, fruits, and leafy vegetables. The dominant food items in Paleolithic era were high plant foods and high animal proteins from lean meat and seafood. Dawn of agriculture was witnessed in Neolithic period. The main food items in this era were grains, legumes, dairy products and vegetable oils. During industrial revolution, the major diets were canned meats, hydrogenated vegetable oils, condensed canned soups and refined grains (*Jew et al*. 2009).

4.3 BIOLOGICAL PERSPECTIVES OF AGING PROCESS AMONG DIFFERENT POPULATIONS

The process of aging is defined as those that increase the susceptibility of individuals, as they grow older, to the factors that eventually lead to death (*Jayanthi et al*. 2010). The academic study of the aging process includes gerontology, the broad study of the aging process, and geriatrics, a speciality concentrating on medical problems associated with growing older. Gerontology utilizes multidisciplinary concepts and approaches in an attempt to understand all aspects of the complex aging process. Three academic areas have contributed substantially to gerontology: biological aging, concerned with longevity and how (and why) the body changes as aging occurs; psychological aging, concerned with adaptive capabilities including memory, intelligence, and how individuals cope with their own aging; and social aging, concerned with social roles and expectations for older adults in a particular culture or society (*Saxon et al*., 2010).

Numerous physical changes historically attributed to aging are now recognized as more likely to be caused by lifestyle variables. For example, aches and pains traditionally attributed to aging are more likely due to a sedentary life style or disuse of abilities rather than to aging per se. Obviously, some factors associated with aging are non-modifiable (i.e., genetics, gender and age), but others can be modifying by lifestyle (i.e., exercise, nutrition, smoking, and stress management). Since a substantial part of the aging process depends on lifestyle, we as individuals can make significant choices to increase the probability of healthy, positive aging. Three lifestyle factors having a major impact on the manner in which we do regular exercise, proper nutrition, and stress management (*Saxon et al*., 2010).

Over the entire span of hominid evolution, life span (the maximum number of years an individual can live) (veryhealthy.com) increased from 53 years in Australopithecines to 122 years in modern humans, whereas life expectancy (an average number of years a person can expect to live) (veryhealthy.com) was 15 years for Australopithecines and 25 for modern human for the recent centuries (Ice, 2003).

In India, the life expectancy was 32 years (https://brainly.in/question/11232730) during the British rule and increased to 69.37 in 2020 (https://www.macrotrends.net/countries/IND/india/life-expectancy). Variation in life expectancy was observed in different countries, the lowest life expectancy (54.36) being in Central African Republic and highest (85.29) in Hong Kong for both sexes. Across the globe, women showed 4.8 years higher life expectancy than men (https://www.worldometers.info/demographics/life-expectancy/). The gender gap in life expectancy at age is larger in regions (Eastern and South East Asia (3.4 years), Latin America and Caribbean (2.8 years) and Europe and North America (3.1 years) with high life expectancy at birth whereas lower gap observed in regions (Sub-Saharan Africa (1.3 years), Central and Southern Asia (1.1 years) and Oceania (0.6 years) with lower life expectancy at birth. It was estimated that women aged ≥65 years will form 54% of population in

2050 and those women aged ≥80 will decline from 60% in 2019 to 59% by the year 2050 (World Population Aging, 2019). Due to HIV/AIDS the decrease in life expectancy for both sexes of 10-20 years was observed in the countries of African continent (https://www.who.int/whr/2004/chapter1/en/index1.html).

Rising life expectancy, decreased fertility and improvements in life expectancy at birth and death is contributing to higher number of older adults. The number of persons aged 65 years was 703 million in 2019 contributing 9% in the world population and their number will be doubled by the year 2050 in Northern Africa, Western Asia, Central and South Asia, Latin America, the Caribbean, Eastern and South-Eastern Asia. The largest number of older adults (260 million or 37%) is represented by regions such as Eastern and South-Eastern Asia and Europe and North-America together contribute 200 million or 28.5%. This trend will be continued in 2050 also for Eastern and South-Eastern Asia. In 2019 one in 11people are aged >65 years and by 2050 one in 6 people will be aged 65 years. Older people in 2019 constituted 1/5th population in 17 countries and they will be reaching 61% in 155 countries by the year 2100. Among countries, Japan is the most aged country and it was estimated this trend will continue in 2050 also (World Population Aging 2019). Changing demographics can be fertile area for human biologists to explore its impact on culture and variation (Ice, 2003).

Older people not only differ with young people but also within and between populations and also between the organ systems of the same individual. Differences in exposure to environmental, economic and social factors were reported to be responsible for varying aging rates among populations. Role of not only evolutionary, genetic, ecological, economic and socio-cultural factors individually needs to be explored but also the interactions of these factors in causing the variation in the process of aging. Biological anthropologists with the emphasis on holistic, evolutionary and cross-cultural research orientation can make significant contribution to the studies on aging (Ice, 2003).

4.4 SUMMARY

Bio-cultural aspect has been intrinsic part of physical/biological anthropology but it gained currency when the Human adaptability project of International Biological Programme was initiated in 1960. Bio-cultural studies employ variables such as biological, cultural (behavioural and psychological) and environmental (biotic (living) and abiotic (non-living)). Bio-cultural studies, explore the relationship between these variables at the population level particularly those contributing to the interaction of biology and culture and also attempt to understand human variation. Bio-cultural approach bridge gap between sub disciplines of biological/physical and social/cultural anthropology and promote integrative thinking. Disease is a pathological state of the organism and considered as western concept. It was observed there are universal perceptions and society specific perceptions with reference to health and disease. Though the concept of health and illness are biological but they are also related with socio-cultural contexts. Certain symptoms were found to be unique to specific cultural group known as culture bound syndromes.

Food consumption ensures availability of essential nutrients to the body and its expenditure provides energy to perform physical, mental and reproductive roles and maintenance of health. Culture facilitates the availability and consumption

of food. Jerome et al. (1980) explained ecological model for food and nutrition and opined that environmental and social factors influence the nutrition of people. The bio-cultural approach emphasizes the investigation of interaction of both sectors. Interaction of biology and culture takes place through genetic, physiological and socio-cultural adaptations. Human diet evolved in four stages. 1. Miocene to early Pleistocene 2. Paleolithic 3.Neolithic and 4. Industrial revolution.

Biological anthropologists developed interest on aging due to the following reasons: paradox phenomenon of aging, availability of little information on the role of ecological and evolutionary factors in causing life span differences among populations, lack of clarity on the role of menopause as adaptive/evolutionary strategy/ by product of modern life, patterns of variations in aging among populations and demographic trends of aging. Role of not only evolutionary, genetic, ecological, economic and socio-cultural factors individually needs to be explored but also the interactions of these factors in causing the variation in the process of aging. Biological anthropologists with the emphasis on holistic, evolutionary and cross-cultural research orientation can make significant contribution to the studies on aging.

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4.6 ANSWERS TO CHECK YOUR PROGRESS

1) It is the scientific exploration of the relationships between human biology and culture. Looking for the biological roots of human behaviour, bio-cultural anthropology attempts to understand how culture affects our biological capacities and limitations.

2) This aberrant behaviour characterized by four phases. First phase or prodrome containing irritation and social withdrawal; second phase involves symptoms like sudden onset of extreme and wild excitement, running in the streets, shouting, tearing off clothes and throwing available objects; third phase consists of fits, stupor, calmness or comma; and final phase is characterized by complete amnesia (Fulk, 2012). This condition is caused by hypervitaminosis A due to higher consumption of liver and fats of arctic and marine animals, which are rich sources of Vitamin A in traditional diet of Eskimo and storage of Vitamin A in poisonous quantities.

3) This condition is due to deficiencies, excesses or imbalances in a person's intake of energy or nutrients.