

Model Answers_AS-2956 (B)
B.A/ BSc. First Semester
(End Semester) Examination, 2013
ANTHROPOLOGY
Paper: Second
(Human Variation)
Time Allowed: Three hours

Maximum Marks: 30

Passing Marks: 12

Section – ‘A’

1x10=10

Note: *Select one of the most appropriate answer from the following objective questions.
Each question carries 1 mark.*

- 1.(i) Hair form is an example of: (a)
- a. **Anthroposcopic variables**
 - b. Anthropometric variables
 - c. Genetic variables
 - d. Special characters
- (ii) ‘*Anthropology on Human Nature, Man’s Most Dangerous Myth*’ was written by (c)
- a. J.B.S. Haldane
 - b. E. Hooton
 - c. **M.F. Ashley Montagu**
 - d. Carleton S. Coon
- (iii) A race is a great division of mankind, the members of which are characterized by a combination of morphological and metrical features derived from common descent, (a)
was given by
- a. **E. Hooton**
 - b. H.H. Risley
 - c. Montagu
 - d. Curt Stern
- (iv) Gregor Mendel’s success was the result, in part, of the following factor (d)
- a. Pairs of sharply contrasting features were used
 - b. Traits were precisely defined
 - c. Results were quantified
 - d. **All of the above**
- (v) Who divided races into geographical, local & micro races? (d)
- a. Hooton
 - b. Brues
 - c. Anders Retzius
 - d. **Garn**
- (vi) Who discovered double helix structure of DNA? (b)
- a. Robert Hook
 - b. **Watson & Crick**
 - c. Hugo de Vries
 - d. None of these
- (vii) Who is considered as father of genetics? (b)
- a. Darwin
 - b. **G.J. Mendel**
 - c. S. C. Roy
 - d. None of the above
- (viii) DNA is a chain of (a)
- a. **Polynucleotide**

- b. Cell
 - c. Chemical
 - d. Micro-organism
- (ix) The inheritance pattern of differing traits are independent of one another. (b)
 This is the principle of
- a. Segregation
 - b. Independent assortment**
 - c. Dominance
 - d. None of the above
- (x) An albino male marries a female known to be a carrier for albinism. What is the probability of their first child being an albino (b)
- a. 0
 - b. $\frac{1}{4}$**
 - c. $\frac{1}{2}$
 - d. $\frac{3}{4}$

Section-'B

4x05=20

Note: Write long answer of the following questions. Attempt any four questions.
 Each question carries 5 marks

2. What is race? Explain the geographical distribution and morphological characteristics of Negroid and Australoid group?

2Answer:

The single human species of *Homo sapiens* exhibits differences in its biological characteristics from one part of the world to another. However, this differentiation occurs only within the possible range of variation in the species-specific characteristics. Several of these variations are prominent and easily identifiable. Several groups of human populations thus differ from one another. Each group of human population that exhibits similarities in its biological that exhibits similarities in its biological characteristics and differs from another group is called a race.

The variations between races are mainly;

1. Morphological
2. Serological
3. Genetical
4. Geographical

Definition of race

Hooton (1926) defined race as a great division of mankind, the members of which, though individually varying are characterized as a group by a certain combination of morphological and metrical features, primarily non-adaptive, which have been derived from their common decent.

The functional significance of the variation tends to be more evident in the quantitative morphological and physiological characteristics than in the qualitative biochemical and serological ones. Because they have been longer known and are more fully and easily studied.

The geographical and morphological characteristics of Negroid and Australoid are as follows,

1.Negroid

Geographical distribution: Represented by the Kadars, Pulayans (Cochin and Travancore, Irular and Primitive tribes of Wynad. They are considered to be autochthones of India.

Morphological Features: Skin Colour – Dark Brown to Dark Black; Hair - Woolly in form; Head Form - Small, Round, Medium or Long; Forehead is Bulbous; Supraorbital Ridges - Smooth; Eyes - Dark in Colour, Nose Form - Straight, Flat and Broad; Stature- Very Short height or Pygmy.

2. Australoid

Geographical distribution: The Australoids are referred also as Proto - Australoid, Pre - Dravidian, Nisada and Veddid. The representatives of this group are some tribes of South India, namely - Urali, Kannikar, Malapantarram, Paniyan, Kadar etc.

Morphological Features: Skin Colour - Dark Complexion; Hair - Dark colour and Wavy in form; Head Form - Long (Dolichocephalic); Eye - Dark in Colour; Nose Form - Broad; Stature – Short height.

Conclusion: The differences exist either in morphology (external appearance) or serology (blood group type) are a product of differences in the genetic makeup of man. A morphological trait or a serological trait or any other trait for that matter is genetically determined and genetically inherited. Thus, it is inbreeding within a race that makes racial characteristics hereditary. Variations between races are a result of mendelian segregation and assortment of genes, non-random mating, mutation, genetical drift, natural selection and geographical isolation.

3. Write short note on

a. Recombinant DNA technology

b. Concept of race

3 a. Answer

Recombinant DNA is what you get when you combine DNA from two different sources.

For example: Mouse + Human DNA

Human + Bacterial DNA

Viral + Bacterial DNA

Human + (other) Human DNA

By this technique, the DNA fragments are generated with the help of restriction endonucleases. These fragments are then incorporated into a suitable vector (plasmid), which in turn is introduced into a host organism (usually bacteria E.Coli). The clones with specific DNA sequence are selected for the analysis of gene structure and diagnosis of genetic disorders either directly or by linkage of both adults and foetus.

The analysis of human genes, particularly those in the beta globin gene region, is made by arranging the fragments generated by various restriction enzymes. In this region there are seven genes, two pseudo genes and E, Gy, Ay, delta and beta. In gorillas, chimpanzees, and Old World monkeys, only five functional genes are found. The haemoglobin changes from the foetal to adult type and the sequence of their formation during human development are similar to the sequence of these genes in beta globin region. Each of the globin genes is about 1.5 kb (1kb= 1000 base pairs) in length and spacer DNA in between them. With the help of suitable probes, the probe in site can be hybridized to locate the gene on a particular region of a specific chromosome. For instance, this technique enabled the location of beta globin genes near the centromere of the short arm of Chromosome 11.

Thus DNA probes are of vital use in this technique. Once the structure of protein or mRNA is known, DNA can be synthesized. Similarly DNA can be located using the DNA probe in the genome of any organism.

Besides the identification of DNA of any micro-organism or virus, DNA probes can also identify mutation or changes in any gene. Every individual DNA has unique hyper-variable regions with the short segments of bases uniquely repeated. If the target DNA is not in sufficient quantity to apply DNA probe, Polymerase Chain Reaction (PCR) technique can be employed for making thousands of copies of a `

3 b. Answer

Definition of race:

Montagu (1942a) defined a race or an ethnic group as representing one of a number of populations comprising the single species *Homo sapiens*, which individually maintain their differences, physical and cultural, by means of isolating mechanisms such as geographic and social barriers.

Concept of race:

The single human species of *Homo sapiens* exhibits differences in its biological characteristics from one part of the world to another. However, this differentiation occurs only within the possible range of variation in the species-specific characteristics. Several of these variations are prominent and easily identifiable. Several groups of human populations thus differ from one another. Each group of human population that exhibits similarities in its biological characteristics and differs from another group is called a race. The variations between races are mainly morphological, serological, genetical and geographical isolation.

Thus population is defined as a group of human population that inbreeds and shares a common gene pool, at the same time it differs in its gene frequencies from other groups. Physical anthropologists and other scholars denounce the very concept of race. Ashley Montague, the chief protagonist has long held the opinion that races are merely products of human imagination. There is no doubt about the fact of human geographical variability, most manifest on a continental basis. The usual division of our species into races or ethnic groups, however, often depends upon a faulty perception of human differences. For example, the size and form of the human face differs considerably throughout the world and the proportions of the lower limbs and the trunk vary over a broad range. Many more subtle differences between human populations, such as those in frequencies of different blood groups, types of blood protein (including enzyme) polymorphisms and DNA markers can be determined only with the help of serological, biochemical and molecular techniques and sophisticated equipments, but they exist nonetheless.

Diverse as they are, these definitions emphasize first an assumption of the role of geographic isolation in race formation. Second, most agree on the importance of breeding population in forming a collection of genes that sets the race apart.

Race is a classification based on traits which are

- a. hereditary
- b. traits transmitted by heredity which characterise all the members of a related group.

4. Explain the phenomenon of Population genetics and Mendelian population.

Answer 4:

Introduction:

The primary aim of physical anthropology is to study human biological evolution in combination with cultural evolution. This study reveals that the living populations in space and time irrespective of their racial affinity, language, and religion have descended from a common ancestor. This reality and the existing human diversity are the result of biogenetic interactions. It is clear that genetics provides a basis for discerning similarities and differences between parents and offspring. The accumulated genetic changes over generations result in evolutionary changes. With the development of the theories of population structure, genetics made a firm foundation for the classification of human variation into clear evolutionary categories with a biological meaning. Human populations or sub-types with the human species are classified into races, ethnic groups, isolates or Mendelian populations on the basis of the difference in the frequencies of the phenotype and genotypic characters besides gross climatic characters.

Phenomenon of Population genetics

The study analysis of population structure of natural populations by the application of Mendelian principles of heredity is known as Population genetics. The study involves the distribution and behavior of genes as well as factors responsible for the maintenance or changes in genes and genotype frequencies and its underlying factors become the basis of evolution. The measurement of genetic composition of populations and their description in qualitative terms by mathematical analysis is carried out by the models and analytical tools of population genetics. The gene frequencies which are calculated by the application of elementary algebra facilitate the description of the genetic structure or composition of a population. The gene frequencies, as measured by observations and theoretical calculations, enable the understanding of differences within subgroups of a species as well as interpretation of their evolutionary status. The evolving populations can be distinguished from the non-evolving ones on the basis of their equilibrium and non-equilibrium estimates.

The concept of population is as much important in modern prehistoric physical anthropology as in the biology of contemporary human groups. The Palaeo-population genetics depend on fragmentary evidence of human remains for the reconstruction of population and as such one has to look for genetically determined characters to understand the population interrelationships rather than depending on measurements. However, population and genetics assume the level of basic conceptions of prehistoric anthropology by which population becomes important in the study of skeletal populations. The importance of genetic component lies in the morphological variations and measurements of size and form to understand the heritability and hierarchy of different variables and indices. It is necessary to understand the role of different processes such as selection, isolation effects, heterosis and assortative matings, genetic drift to understand the change in the genetic structure of population. For this purpose, polyfactorial characters of stature and other measurements in the study of breeding depression and heterosis etc have to be undertaken to understand and explain the morphological differentiations.

Phenomenon of Mendelian population

According to Theodosius Dobzhansky, a Mendelian population is 'a reproductive community of sexual and cross fertilizing individuals which share in a common gene pool'. A mendelian population may contain a number of subunits which are by themselves smaller Mendelian populations. A species is the largest Mendelian population. Species distinguish themselves from each other as they do not share the same gene pool. Such systems are known as closed genetic systems. The human races which differ from each other are essentially Mendelian populations of the species *Homo sapiens*. This is so because their gene pools to some extent differ from each other and such differences are usually manifested phenotypically. However, these racial groups are not closed to the exchange of genes with each other and as such they have the potential and facilitate actual

gene flow between their respective gene pools. It is important to note in this connection that all Mendelian populations are nothing but 'breeding isolates'. Thus a breeding isolate is a population whose members find most of their mates within their respective groups. Breeding isolation occurs mostly due to geographical barriers besides the religious, social and psychological factors. If the breeding isolation is prolonged, it results in the development of differences in the genetic makeup between human population groups. This speaks of the differences in the gene frequencies between these breeding isolates.

5. What is Pedigree analysis? Explain the construction of pedigree symbols?

Answer 5:

Pedigree analysis is one of the chief methods of genetic study of man employed by physical anthropologists as well as human geneticists. The method implies the observation of the patterns of distribution of genetic traits in kindreds also called pedigree patterns. For this purpose the investigator has to necessarily depend on information concerning the family history to get at the genetics of a given trait. Once the family history is known, the pedigree becomes a short-hand or graphic representation of the details. The characteristic features of the individuals in families range according to sex, generation and biological relationship to each other. It is through pedigree patterns that we can understand the mechanism of Mendelian Principles of segregation and independent assortment as well on allelism and linkage. Both single factor inheritance and polygenic inheritance, as in the case of intelligence by the analysis of intrafamilial similarity, can be studied by this method.

The method of pedigree analysis, considering phenotypic differences between two individuals regards them, one as normal and the other as abnormal or both as variants of normal and abnormal traits. Many abnormalities can be genetically distinguished from the normal persons by a difference at a single pair of loci while in the case of normal variants the genetic differences between persons with a trait are often polygenic or due to difference at many pairs of loci.

Whenever an investigation is undertaken, it should be ensured that the investigator is thoroughly acquainted with the field area, the type of instructions to be given to the individuals and the type of information expected, and the reasons for collecting the information from the individuals of study. The next step would be to prepare a key for information on the symbols to be used with clear definitions and to record the prevailing conditions within the family of study. Finally, the information on the pedigrees collected with the help of the informant would be of relevance and purpose to later studies.

The recording of pedigree is done from the concerned person irrespective of whether they seek genetic counseling or not, duly supplemented by other members of the family as well as documents, if any, preserved by them. The details of the information have to be taken not only on the living and affected individuals in the family but also about miscarriages, still births, deceased individuals, and children, if any, spared for adoption. These details are necessary to understand the prevailing situation concerning the lethality or reproductive losses. Other types of information to be collected include the birth place of the individuals in the pedigree makes its analysis possible and also we can have a clearcut idea of the patterns of inheritance and the differential expression in one sex.

There are internationally accepted conventions followed in the proper construction of pedigree charts. The resumption of the study of a particular trait in a family generally starts with an affected person. Such person is referred to as Proband, or as the Propositus for male and Proposita for female, or the index case. Squares and circle symbols are used for males and females. While preparing the pedigree charts the usual practice followed is to place the male first on the left side and the females on the right side joined by a horizontal marriage line, followed by the symbols of the offspring in horizontal row below a line which is connected by vertical lines. Consanguineous marriage is indicated by double marriage lines as $\square = \bigcirc$

which are different from monozygotic pairs. The unlike-sexed twins of all dizygotic twins may be derived as under:

$$\frac{\text{Unlike – sexed DZ}}{\text{All DZ}} = \frac{2pq}{p^2 + 2pq + q^2} = \frac{2pq}{1}$$

Then, all DZ = Unlike – sexed DZ

$$\frac{\quad}{2pq}$$

And all MZ = Total twins – all DZ.

In the diagnosis of zygosity of the twins, two methods are applied to determine whether a particular pair of twins as DZ or MZ. One of these methods is meant to examine the components of the afterbirth of twins. The second method also called as similarity method, determines the degrees of resemblance in physical traits between the twins.

Answer 6b:

Biochemical method:

Human biochemical investigations have resulted in the development of a good deal of procedural methods. At least the following procedures play an important role in the biochemical studies.

A. Sequencing of DNA

B. Southern, Northern and Western Blotting

A. Sequencing of DNA:

The sequencing of DNA is done by Maxam Gilbert method, as a result of which the DNA is cleaved by four restriction enzymes which cut DNA at different bases. The four sets of samples, each with differing length of DNA fragments, represent the position of the bases. The electrophoresis of these four samples is done by four parallel gels which indicate the position of the bases.

B. Southern, Northern and Western Blotting:

This method derives its name from the techniques developed respectively for the presence of specific DNA, RNA and protein in the order of Southern, Northern and Western blotting. The first of these techniques, namely Southern blotting was described for DNA followed by the other techniques for the presence of RNA and proteins in a similar way. In the Southern blotting, the restriction enzyme cleaves the DNA and the fragments are separated by gel electrophoresis. The blotting of fragments is done on nitrocellulose filter paper and hybridization by labeled probe. In like manner, the RNA molecules are detected by the Northern blotting technique. In the case of proteins where the Western blotting technique is used, electrophoresis is employed for separation of proteins followed by their blotting on nitrocellulose filter. The substance is then treated with the antibody resulting in antigen-antibody. This complex is reached with labeled antibody the excess of which is washed out. The presence of such labelled antibody is done by Auto-radiography.

7. Evaluate different methods that are employed in studying Human genetics?

Answer 7:

The different methods employed for the study of human genetics are

- a. Pedigree analysis,
- b. Twin method,
- c. Co-twin method,
- d. Family studies,
- e. Biochemical methods,
- f. Chromosomal analysis,
- g. Immunological methods,
- h. Recombinant DNA technology,
- i. Site directed Mutagenesis
- j. Foetal DNA

a. Pedigree analysis:

Pedigree analysis is one of the chief methods of genetic study of man employed by physical anthropologists as well as human geneticists. The term pedigree is used by breeders to indicate the quality or type of genetic background that a particular animal has family histories of traits, to study human conditions. A deceased individual is indicated in a pedigree by a slash through the icon. Pedigrees involving divorces, remarriages, and adoptions are more difficult to notate, as well as to analyze, conclusions must be drawn carefully. In human pedigree analysis, the trait of interest (usually a “condition”) is indicated with a shaded icon, whether it happens to be recessive or dominant. All genetic disorders are fairly rare, some are recessive such as Phenylketonuria, sickle cell disease, and cystic fibrosis and some are dominant such as polydactyly, the Marfan syndrome, and Huntington’s disease. Recessive conditions can be carried in a family for many generations without anyone being aware of them until a carrier has children with another carrier. In other words, they can skip one or more generations, just as the recessive pea plant traits identified by Mendel do.

The rules for constructing Pedigrees may include the following:

1. Pedigrees always have a title.
2. Pedigrees always have a key.
3. Females are represented by circles.
4. Males are represented by squares.
5. The affected individual is represented by another color.
6. Members of the same generation are at the same horizontal level.
7. Parents of offspring are connected with a horizontal line.
8. Offspring are connected to their parents with a vertical line.

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pair of loci while in the case of normal variants the genetic differences between persons with a trait are often polygenic or due to difference at many pairs of loci.

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The various symbols for pedigree charts dealt with various specifications were adopted internationally by the Federation of Eugenic Organizations for research in human heredity. The pedigrees although contain all the information regarding the manner of inheritance of the concerned gene or genes, it may be conclusive unless it is very large or the gene shows cent percent penetrance. Whenever the pedigrees are very short, it is necessary to compare several of them to come out with an objective interpretation.

d.Family Studies:

Genetic investigation of families assumes significance as a method for the study of man because the families are represented by parents each of whom passes half of its genes to an offspring and accordingly one-half of the common genes each are present in the parent and offspring. This half known by the term 'coefficient of relationship' exists between parent and the offspring. In the same manner sibs also have half 'coefficient of relationship' due to the fact that if a gene is inherited by one sib, another sib has 50% chance of inheriting the same gene. So also the grand children of the next generation inherit one half of a gene from parents.

Accordingly the likelihood of a gene transmitted by grandparent to grandchildren is $\frac{1}{2} \times \frac{1}{2} = \frac{1}{4}$ which is the coefficient of relationship between them. Such a coefficient of relationship exists between uncle-aunt, and nephew-niece. The coefficient of relationship between first cousins can be expressed as $\frac{1}{2} \times \frac{1}{2} \times \frac{1}{2} = \frac{1}{8}$.

This kind of coefficient of relationship is known as additive genetic system by which an individual is like half of its parent, a quarter like its grand parent, a quarter like its uncle, maternal aunt and 1/8 like a first cousin. Thus the family studies helps in understanding the genetic relationship through the inheritance of factors that have additive genetic basis, environmental component and a dominance factor.

8. Briefly explain the methods of studying Chromosomal disorders?

Answer 8:

The patients with chromosomal abnormalities of suspected chromosomal aberrations can be studied with the help of three different laboratory tests. One of them is meant to analyze mature neutrophils for the presence of a nuclear appendage, the drumstick. By the second test the cells are studied for the presence of sex chromatin bodies (Barr bodies) in their nuclei. The third test is carried out for counting the chromosomes in the cell. The first two tests are usually carried out as screening tests and are relatively less expensive, while the test on chromosome count is expensive which is though a difficult one is reliable.

In the case of the first test the peripheral blood smear when stained present mature neutrophils which may have a nuclear appendage called drumstick. The drumstick is a solid, round, discrete head, joined by a single thin strand of chromatin to a lobe of neutrophil. It is about 1.5 μ in diameter and can be seen under microscope at a magnification of X 90, representing sex chromatin. The normal females have drumsticks to the extent of 1-3 percent whereas in the case of normal males there are no drumsticks present. This test should be done by counting at least 200 neutrophils.

In the test on sex chromatin analysis staining has to be done properly for identification of sex chromatin (barr) body as a solid well defined plano-convex mass of approximately one μ in diameter. It lies near or at the inner surface of the nuclear membrane. For this study the specimens from the desquamated buccal, vaginal and amniotic cells comprise the most frequent source. The sex chromatin body represented as the heterochromatic X chromosome is found at the rate of one less than the number of X chromosomes in a cell. In females the incidence varies from 40-60 percent and they are chromatin-positive. Males on the other hand are chromatin-negative due to the absence of these bodies; sex chromatin can be seen in the trophoblast on the 12th day and in the embryo on the 16th day. With the help of sex chromatin analysis from amniotic cells, the sex of the unborn can be predicted.

In the chromosomal analysis test, the cells are grown from a biopsy in tissue culture inhabiting them at mitotic metaphase stage, chemically followed by sorting and counting the chromosomes. The specimens for the test are usually taken from peripheral blood, bone marrow, skin and testes. The tests should be performed on a significant number of cells. The analysis is also called a karyotype which consists of a systematic number of chromosomes of a single cell.

The well-known chromosomal disorders are

- a. Klinefelter syndrome,
- b. Turner's syndrome,
- c. Down's syndrome or Mongoloid idiocy,
- d. Cri du Chat or Cat Cry syndrome,
- e. Philadelphia syndrome,
- f. Patau's syndrome,
- g. Edward's syndrome
- h. Wolf-Hirschhorn syndrome
- i. Robertsonian chromosome or D/D Translocation syndrome,
- j. The Laurence Moon-Biedl syndrome,
- k. Adrenogenital syndrome
- l. Festicular Feminization syndrome,
- m. Mosaic Trisomy 16

For Example:

Klinefelter syndrome was discovered in 1959 by Jacob and Strong. The phenotypic sex of the affected individuals is somewhat confused due to the disturbance of the development of primary and secondary sex characters. Phenotypically such persons appear like a 'male' with a chromosomal constitution of XXY gonosomes, the total number of chromosomes being 47. Such abnormal conditions as these that have been challenging to geneticists found clarification as also identification of sex after the discovery of the chromatin or the barr body on the inside of the female nuclear membrane by Murray L. Barr and Bertram of the University of Western Ontario and others.

Klinefelter syndrome caused by nondisjunction is a good example of involvement of sex chromosome in aneuploidy. Its incidence is approximately 1 in 700 births of male children. The infants may appear to be normal. In course of maturity they develop certain phenotypic features which suggest that they have 47 chromosomes instead of 46, the extra chromosome being an X chromosome. The individuals are definitely males, but their penis and testes are small, and they have somewhat enlarged breasts (a condition called gynecomastia) and sparse body hairs. They are usually always sterile. Some of them are also mentally retarded. However, some have normal intelligence. For normal male sex development one Y chromosome is needed. In an affected Klinefelter individual, apparently the Y chromosome behaves normally. But the presence of an extra X chromosome produces more female sex hormone and thereby the balance is disturbed effecting the development and maturation of the normal male characters. Their limbs become long in proportion to the rest of the body. Most of the Klinefelter individuals are sex chromatin positive.