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

Maximum Marks: 30 ***Passing Marks: 12***

Note: *Attempt questions of all **two** sections as directed. Distribution of marks is given with sections.*

Section – 'A' **1x10=10**

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

1. (i) d. All of the above
   (ii) b. Bred only with the same kind and shows the same trait or traits over many generations
   (iii) d. All of the above
   (iv) d. All of the above
   (v) b
   (vi) b. Recessive
   (vii) b. Phenotype
   (viiii) b. Multiple allele
   (ix) b. Autosomal recessive
   (x) a. Anthroposcopic variables

Section-'B **4x 5=20**

Note: *Write long answer of the following questions. Attempt any four questions. Each question carries 05 marks.*

2. What is Human variation? Explain the genetic basis of race?
Answer 2:

Human Variation:
The single human species of Homosapiens 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

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 are known and are more fully and easily studied. 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.

Genetic basis of race

The relationship between race and genetics is relevant to the controversy concerning race. In everyday life many societies classify populations into groups based on phenotypical traits and impressions of probable geographic ancestry and socio-economic status - these are the groups we tend to call "races". Because the patterns of variation of human genetic traits are clinal, with a gradual change in trait frequency between population groups, it is possible to statistically correlate groups of physical traits with individual geographic ancestry. This is due to endogamy within kin groups and lineages or national, cultural or linguistic boundaries. This causes genetic groups to correlate statistically with population groups when a number of alleles are evaluated.

If a person has light skin, light hair and blue eyes, a combination of traits that seems to have evolved in Northern Europe and is found at a high frequency there, it is probable that person has some recent European ancestry. In a similar way, genetic analysis enables us to determine the geographic ancestry of a person pinpointing the migrational history of a person's ancestors with a high degree of accuracy, and by inference the probable racial category into which they will be classified in a given society. In that way there is a distinct statistical correlation between gene frequencies and racial categories. However, because all populations are genetically diverse, and because there is a complex relation between ancestry, genetic makeup and phenotype, and because racial categories are based on subjective evaluations of the traits, it is not the case that there are any specific genes that can be used to determine a person's race.

Research in genetics offers a means to classify humans which is more precise than broad phenotypically based racial categories, given that genetics can provide a much more complex analysis of individual genetic makeup and geographic ancestry, than self-identified membership of a racial category. With a blood transfusion, for example, it is vital to know the genetically determined blood type of the donor and recipient, but it is not helpful to know their respective geographic ancestries. Most physical anthropologists consider race to be primarily a social category that does not correspond significantly with biological variation, but some anthropologists, particularly forensic anthropologists, consider race a useful biological category. They argue that it is possible to determine race from physical remains with a reasonable degree of certainty; what is identified is the geographic phenotype.

3. Write short note on

a. Caucasoid b. Mendel Law of Segregation

Answer 3a:

Caucasoid is one of the major races in the World and in total there are 12 sub races of Caucasoid. Caucasoid possesses dolicocephalic head and also contains brachycephaly and mesocephaly in considerable numbers.

The characteristic features of the Caucasoid race

Characters	Type(s)	Caucasoid
Head form		Long to broad
Head Height		Medium to very high
Face form		Narrow to medium broad
Prognathism		Absent
Nose	Form	Long and narrow
	Bridge	High
	Profile	Straight, concave or convex
Eye	Form	Occasional presence of lateral fold
	Colour	Light blue to dark brown
Hair	Form (Head)	Straight to wavy
	Colour	Light blue to dark brown
	Texture	Fine to medium
	Cross-section	Usually ovoid
Body hair		Moderate to heavy
Beard and Moustache		Moderate to heavy
Lips		Thin to medium
Skin colour		Reddish white to light brown
Stature		Medium to tall

Answer 3b:
Mendel law of Segregation:

The principles that govern heredity were discovered by a monk named Gregor Mendel in the 1860's. After completing his experiment on garden pea in 1866, Gregor John Mendel published his important conclusions in regard to heredity in the Proceedings of the Natural History Society of Brunn. But unfortunately the scientist did not pay much attention to his findings and thus his result remained unnoticed until 1900. About the beginning of the 20^{th} century, 16 years after Mendel's death, his laws were rediscovered by three European scientists, Correns, De Vries and Von Tshermark. The rediscovery created a sensation all over the scientific world. Mendel's theory brought a dramatic change in the thinking about man and heredity. His basic rules of heredity are applicable to all forms of life.
Mendel's law of segregation is also known as Law of Purity of Gametes. When a pair of allele are brought together having contrasting characters remain together without mixing and separate from each other when the hybrid forms gametes.

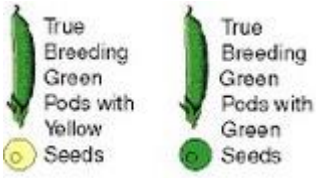
There are four main concepts related to this principle. They are as follows:

- A gene can exist in more than one form.
- Organisms inherit two alleles for each trait.
- When gametes are produced (by meiosis), allele pairs separate leaving each cell with a single allele for each trait.
- When the two alleles of a pair are different, one is dominant and the other is recessive.

For Example:

The gene for seed color in pea plants exists in two forms. There is one form or allele for yellow seed color (Y) and another for green seed color (y). In this example, the allele for yellow seed color is dominant and the allele for green seed color is recessive. When the alleles of a pair are different (heterozygous), the dominant allele trait is expressed and the recessive allele trait is masked. Seeds with the genetic makeup of (YY) or (Yy) are yellow, while seeds that are (yy) are green.

Law of Segregation



4. Distinguish between Mongoloid and Negroid morphological features and explain the geographical distribution of these races?

Answer 4:

Mongoloid race originated in the vast steppe lands of Central Asia from where they have moved to the different parts of the world. They are characterized by yellow or yellow brown skin colour, straight hair form, usually brachycephalic headform, very low nasal root, low and medium broad nasal bridge, usually concave or straight nasal profile, oblique with narrow slit like opening of eyeform and total epicanthic fold in the upper lid, medium to dark brown eye colour, flat face form and variable stature. This race is divided into four sub-races.

1. Classic Mongol
2. Arctic Mongol
3. Indonesian Mongol
4. American Mongol

Negroid race is characterized by dark brown to dark skin colour, woolly or frizzly hairform, black hair colour, little body hair, and sparse face hair, dark brown to dark eye colour, usually dolicocephalic headform, low and broad nasal bridge, broad and flat nose form, rounded forehead, small brow ridges, thick and everted lips, round and receding chin and predominantly short, wide ears with rolled helix and little or no lobe. There are five subraces recognized in the Negroids,

1. Pygmy Negritos,
2. Forest Negroes,
3. Nilotic Negroes
4. Oceanic Negroes
5. Bushmen-Hottenhot

Characters	Type(s)	Mongoloid	Negroid
Head form		Broad	Long
Head Height		Medium	Low to medium
Face form		Medium to very broad	Medium broad to narrow
Prognathism		Rare	Marked
Nose	Form	Medium	Broad
	Bridge	Low	Low
	Profile	Usually concave	Straight or Concave
Eye	Form	Medial epicanthic fold	Vertical fold common
	Colour	Brown to black brown	Dark brown to black
Hair	Form (Head)	Straight	Woolly
	Colour	Dark brown	Dark brown to black
	Texture	Coarse	Coarse
	Cross-section	Round	Flat oval
Body hair		Sparse	slight
Beard and Moustache		Slight to moderate	Absent to slight
Lips		Medium	Very thick
Skin colour		Light yellow to yellowish brown	Brown to sooty black
Stature		Medium short to medium tall	Very short to tall

5.What is RFLP? How does RLFP is useful in the light of Anthropological studies?

Answer 5:
Restriction Fragment Length Polymorphism:

Restriction Fragment Length Polymorphism is one of the genetic marker. A genetic marker is a gene or DNA sequence with a known location on a chromosome that can be used to identify individuals or species. It can be described as a variation (which may arise due to mutation or alteration in the genomic loci) that can be observed. A genetic marker may be a short DNA sequence, such as a sequence surrounding a single base-pair change (single nucleotide polymorphism, SNP), or a long one, like minisatellites.

For many years, gene mapping was limited in most organisms by traditional genetic markers which include genes that encode easily observable characteristics such as blood types or seed shapes. The insufficient number of these types of characteristics in several organisms limited the mapping efforts that could be done.

Some commonly used types of genetic markers are

- RFLP (or Restriction fragment length polymorphism)
- SSLP (or Simple sequence length polymorphism)

- AFLP (or Amplified fragment length polymorphism)
- RAPD (or Random amplification of polymorphic DNA)
- VNTR (or Variable number tandem repeat) etc.,

Some of the techniques developed for DNA manipulation are used to detect DNA variations known as **restriction fragment length polymorphisms** (**RFLP**s).

A. Some one per thousand base pairs (bp = nucleotide pairs) varies in the population, i.e. instead of an AT pair, there might be a TA, GC, or CG pair. Often these are polymorphic.

B. Some of these variants involve a sequence susceptible to attack by a restriction enzyme. The DNA of one sequence may be cut by a certain restriction enzyme; the other sequence is not a target for this restriction enzyme and is not cut. Since there will be flanking restriction sites, the first form will generate two DNA fragments; the second only one.

C. The fragments can be separated on the basis of size by means of electrophoresis and detected in Southern blots.

D. The "trait" that is expressed is variation in length of fragments of DNA (restriction fragments); thus the name restriction fragment length polymorphism. Most restriction sites are not polymorphic, however.

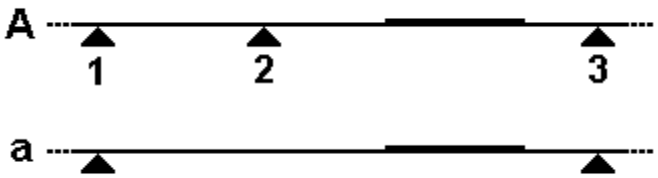
E. These DNA variations are transmitted strictly by Mendelian rules as codominant traits and make excellent genetic markers.

In molecular biology, **restriction fragment length polymorphism**, or RFLP (commonly pronounced “rif-lip”), is a technique that exploits variations in homologous DNA sequences. It refers to a difference between samples of homologous DNA molecules that come from differing locations of restriction enzyme sites, and to a related laboratory technique by which these segments can be illustrated. In **RFLP analysis**, the DNA sample is broken into pieces (digested) by restriction enzymes and the resulting *restriction fragments* are separated according to their lengths by gel electrophoresis. Although now largely obsolete due to the rise of inexpensive DNA sequencing technologies, RFLP analysis was the first DNA profiling technique inexpensive enough to see widespread application. In addition to genetic fingerprinting, RFLP was an important tool in genome mapping, localization of genes for genetic disorders, determination of risk for disease, and paternity testing.

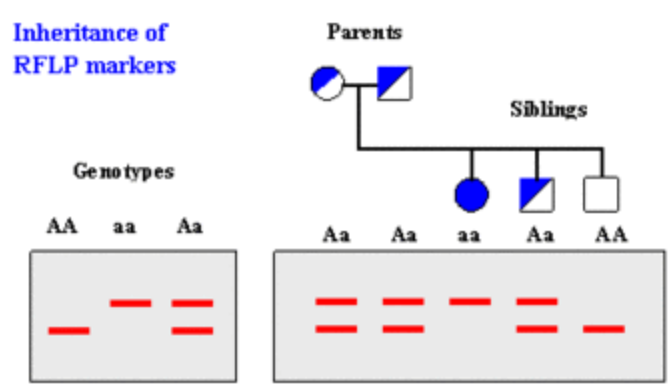
The basic technique for detecting RFLPs involves fragmenting a sample of DNA by a restriction enzyme, which can recognize and cut DNA wherever a specific short sequence occurs, in a process known as a restriction digest. The resulting DNA fragments are then separated by length through a process known as agarose gel electrophoresis,

and transferred to a membrane via the Southern blot procedure. Hybridization of the membrane to a labeled DNA probe then determines the length of the fragments which are complementary to the probe. An RFLP occurs when the length of a detected fragment varies between individuals. Each fragment length is considered an allele, and can be used in genetic analysis.

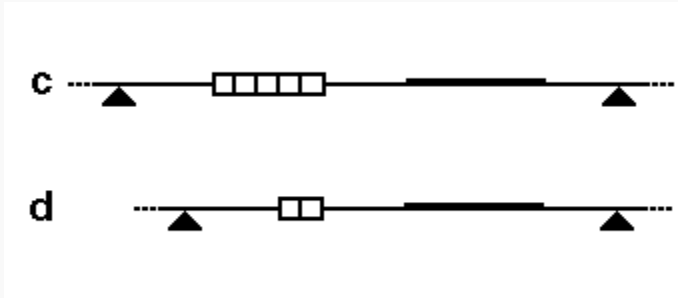
- RFLP analysis may be subdivided into single- (SLP) and multi-locus probe (MLP) paradigms. Usually, the SLP method is preferred over MLP because it is more sensitive, easier to interpret and capable of analyzing mixed-DNA samples. Moreover, data can be generated even when the DNA is degraded (e.g. when it is found in bone remains.)



- Schematic for RFLP by cleavage site loss.



Analysis and inheritance of allelic RFLP fragments (NIH).



Schematic for RFLP by VNTR length variation.

6. Write short note on

a. Genetic polymorphism b. Rh factor

Answer 6a:
Genetic Polymorphism:

Genetic polymorphism refers to a condition under which exist two or more genetically different classes in the same interbreeding population at the same time in the same habitat. Between the frequencies of these alleles an intermediate equilibrium is established and thus the two classes are maintained together in the same population. This is called balanced polymorphism. The two types are retained at equilibrium sometimes due to heterozygote superiority. Sickle cell anaemia is cited as the best example of balanced polymorphism in Homosapiens. The heterozygote which is resistant to malaria maintains its superiority over the normal homozygote with two alleles of haemoglobin, normal and sickling.

According to Fisher's mathematical derivation an equilibrium will be established in case the heterozygote has a higher selective value than both homozygotes. The determination of the frequency of the heterozygote depends on its fitness as compared to those of the homozygotes. It is the environment which determines such a polymorphism which will not be affected by mutations of either of the alleles.

The ABO blood alleles are present in different frequencies in all geographical regions of the world. These alleles vary in their distribution in different ways. These blood types are polymorphic in nature and their diversity is maintained by a selective balance, partly by heterozygous advantage. Though these blood groups have no effect on the bodily structure, they are found to be associated with their proneness to specific diseases. For instance, O group individuals show increased tendency to duodenal ulcers. They also maintain differential foetal mortality in populations. The phenomenon of polymorphism is also maintained by a number of animal and plant species such as common snail with different shell colors, primula plant species with its heterostlyly, moths with melanisms, butterflies with mimicry, chromosomal polymorphism in a species of prosophina.

Answer 6b:
Rh factor:

In 1940 Landsteiner and Weiner demonstrated that a serum may be obtained from a rabbit when the blood of a rhesus monkey is injected into the former. That serum agglutinates blood of certain human individuals and not of others. This new agglutinates factor is called the Rh factor the Rh symbol being obviously derived from the word rhesus. Those persons who possess this factor are called Rh-positive and those who lack it are said to be Rh-negative. This Rh system is quite independent of the ABO blood group and MNSs blood systems.

The Rh (Rhesus) blood group system (including the Rh factor) is one of thirty-two current human blood group systems. Clinically, it is the most important blood group system after ABO. At present, the Rh blood group system consists of 50 defined blood-group antigens, among which the five antigens D, C, c, E, and e are the most important. The commonly used

terms Rh factor, Rh positive and Rh negative refer to the D antigen only. Besides its role in blood transfusion, the Rh blood group system—specifically, the D antigen—is used to determine the risk of hemolytic disease of the newborn as prevention is key.

Rh incompatibility is a condition that occurs during pregnancy if a woman has Rh-negative blood and the baby has Rh-positive blood. "Rh-negative" and "Rh-positive" refer to whether the blood has Rh factor. Rh factor is a protein on red blood cells. If a person have Rh factor, then Rh-positive and if a person doesn't have Rh factor, then they belong to Rh-negative. Rh factor is inherited (passed from parents to children through the genes). Most people are Rh-positive.

For example: A severe hemolytic disease known as erythroblastosis fetalis, is a condition apparent at birth may occur because of Rh incompatibility. This may happen when a Rh-negative mother carries a Rh-positive fetus by her marriage with a Rh-positive male. The Rh antigen from the Rh-positive fetus may penetrate the placenta to enter into the blood of the mother, and thereby cause production of an antibody. As the Rh-negative mother does contain the antigen, this antibody cannot do any harm to the blood cells of the mother. But when this antibody makes its way through the placenta to the Rh-positive fetus, reaction takes place resulting in Erythroblastosis fetalis.

7.What are Mendelian principles and explain each by giving suitable examples?
Answer 7:

A good understanding of the principles of Mendelian inheritance is a prerequisite to the conceptual understanding of evolutionary theory. Indeed, though Darwin himself subscribed to the "blending" theory of inheritance, it has since been determined that evolution by natural selection requires discrete genes.

Mendel experiments:

Gregor Mendel, the Austrian monk famous for his experiments with pea plant characteristics, was the first to identify discrete units of heredity and thus discredit the blending theory. Mendel used characteristics of pea plants and four o'clock flowers to analyze the hereditary patterns of these traits. His historic experiments led him to the conclusion that inherited characteristics were carried in discrete, independent units (later named genes). In Mendel's interpretation, hereditary characteristics occurred in pairs of factors that had specific relationships. Mendel devised two fundamental principles of inheritance:

- **Mendel's Principle of Segregation:** The factors of inheritance (genes) normally are paired, but are separated or segregated in the formation of gametes (eggs and sperm).
- **Mendel's Principle of Independent Assortment:** Each factor's distribution in the gametes is not related to the distribution of any other factor. (This principle is not strictly true due to the organization of genes on chromosomes.)

Mendel also defined and described the relationships between the different factors of inheritance and their effects on the observed characteristics of the organism.

Mendel's Observations:

Mendel made numerous important observations in his exhaustive study of pea plants' characteristics. He elaborated an important distinction between dominant and recessive traits through his work with pea plants.

By studying the seven characteristics of pea plants, such as their

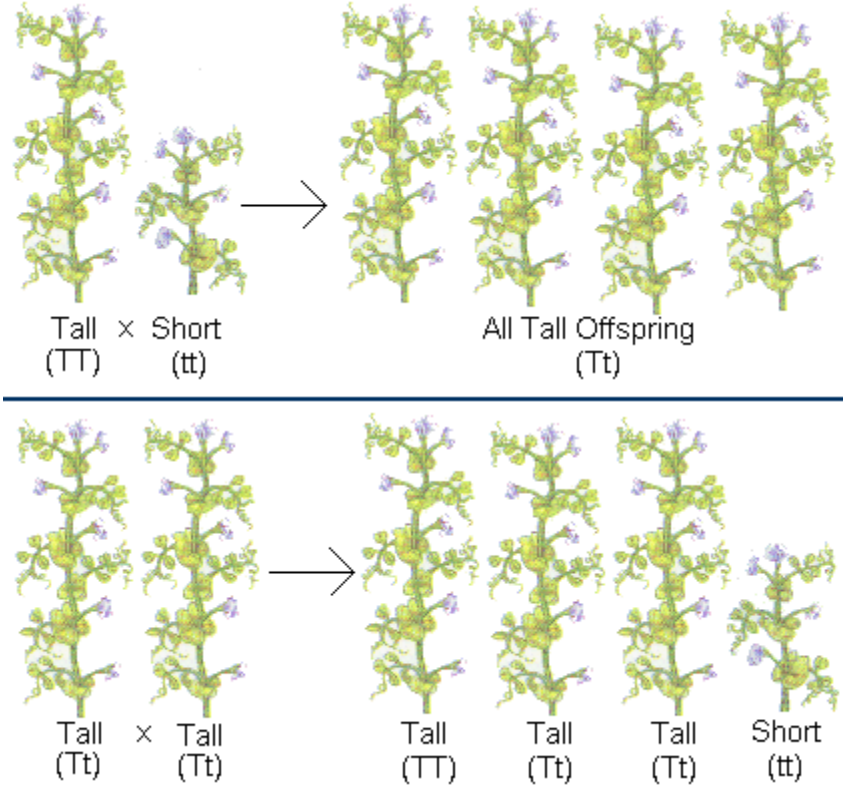
Height – Tall or Short,

Seed shape – Round or Wrinkled,

Seed color – Yellow or Green,

Flower position – Axial or Terminal,

and other traits. Mendel first crossbred one tall, true-breeding plant with one short, true-breeding plant. Contrary to the blending theory, all the offspring were tall. In terms of genotype, the original tall plant was TT (two dominant alleles; homozygous), the short plant was tt (two recessive alleles; homozygous), and the second-generation plants were Tt (one dominant and one recessive allele; heterozygous).



When Mendel next allowed these plants to self-fertilize, he found that the short trait reappeared in the third generation. The ratio of short to tall plants was almost exactly 3:1. Their genotypes were as follows - 1 short (tt) : 2 tall (Tt) : 1 tall (TT).

Phenotypic ratio—3:1
Genotypic ratio—1:2:1
Note: We can also show the same self-fertilization by using Punnet Squares.

8. Explain the approaches used for diagnosis of genetic disorders?
Answer 8:

There are two approaches used for the diagnosis of genetic disorders:

1. Direct method
2. Indirect method

1.Direct method:

By this method, the DNA probes constructed for normal genes are used to probe abnormal genes without strict hybridization. The method has been used to detect sickle cell gene. The direct method is called so because DNA is directly analyzed without cleaving with restriction enzymes. This method also has been used in the detection of point mutations like the alpha anti-trypsin gene. The method is also known as 'Forward Genetics'.

2.Indirect method:

This method is entirely reverse to the direct method and hence known as 'Reverse Genetics'. This is applied in situations where a definite protein or mRNA is not known in the case of a genetic abnormality. Therefore effort has to be made to directly find out deviations in DNA, locate the defect and then place the defective protein. Thus tracing variability in the DNA and linking it with the disease locus have to be done. The study of genetic structure of populations as in the case of blood group studies is carried out by the approach of restriction fragment length polymorphism (RFLP). This is done by cleaving DNA with restriction enzyme, which cuts at definite base sequence resulting in different lengths of DNA. It is believed that such variations in bases arose due to point mutations, small deletion or addition. Such of these restriction fragments of enzymes alter in their mobility on gel electrophoresis. The rate of migration of the fragments in electrical field depends on their size. These RFLPs can be studied directly for deletion detection or for allele linked, locus linked or mutation-specific changes.

For example, the allele-linked RFLP is so called as it denotes common origin of the fragment and allele in the population. In Afro-Caribbean Blacks, HPa1 restriction enzyme has been found to generate 7.0 or 7-6 kb long fragments containing beta globin gene unlike the occurrence of 13.0 kb sickle cell globin gene.