This question paper contains **3** printed pages]

Roll No. | | | | | | | | | | |

S. No. of Question Paper : **5544**

Unique Paper Code : **3182013602**

Name of the Paper : **Human Genetics**

Name of the Course : **B.Sc. (Hons) Biomedical Science (NEP)**

Semester : **VI**

Duration : **3 Hours** Maximum Marks : **90**

(Write your Roll No. on the top immediately on receipt of this question paper.)

Attempt *five* questions in all.

Question No. 1 is compulsory.

Subparts of the questions should be attempted together.

Give illustrations and examples wherever required.

Use of non-programmable scientific calculators is allowed.

1. (*a*) Briefly explain the following (any *three*) : 3×2=6

   (*i*) Microsatellites

   (*ii*) Variable expressivity

   (*iii*) Adoption study

   (*iv*) Epigenetics.

P.T.O.

(*b*) Differentiate between the following (any *two*) : 2×4=8

(*i*) Genotype and Haplotype

(*ii*) Locus heterogeneity and Allelic heterogeneity

(*iii*) Sickle cell anemia and Thalassemia

(*iv*) Aneuploidy and Polyploidy.

(*c*) Expand the following (any *four*) : 4×1=4

(*i*) CFTR

(*ii*) LINE

(*iii*) PAC

(*iv*) cDNA

(*v*) NGS.

2. (*a*) What were the contributions of Galton and Garrod in the field of genetics ?

(*b*) What are genetic markers ? Discuss any *one* genotyping method for each of the two types of genetic markers commonly used. 6,12

3. (*a*) What is sex-linked dominant inheritance ? How is it different from sex-limited and sex-influenced inheritance ? Explain using hypothetical pedigrees for each of the cases.

(*b*) What is a quadruple marker test ? How is it beneficial over the triple marker test in prenatal disease diagnosis ? 9,9

4. (*a*) In a population, two linked loci, A and B, have two alleles each (A/a and B/b). The following haplotype frequencies have been observed :

AB = 0.40, Ab = 0.10, aB = 0.20, ab = 0.30

(*i*) Calculate the allele frequencies for A and B

(*ii*) Determine the linkage disequilibrium coefficient (D)

(*iii*) Compute the normalized linkage disequilibrium coefficient (D')

(*iv*) Interpret your results.

(*b*) What were the goals of the human genome project ? What are the key ethical concerns associated with the Human Genome Project ? 10,8

5. (*a*) How has technology advancement improved the Sanger's method of DNA sequencing ? Support your answer with relevant diagrams and illustrations.

(*b*) Explain how uniparental disomy can lead to genetic disorders despite the absence of chromosomal deletions or duplications. 9,9

6. Write short notes on the following (any *three*) : 3×6=18

(*i*) Hardy Weinberg equilibrium

(*ii*) Genetic counselling

(*iii*) Genetic basis of color blindness

(*iv*) Positional cloning approach for gene identification.