

[This question paper contains 2 printed pages.]

Your Roll No.....

Sr. No. of Question Paper : 1742

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Unique Paper Code : 3182013602

Name of the Paper : Human Genetics (NEP)

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

Semester : VI

Duration : 3 Hours

Maximum Marks : 90

Instructions for Candidates

1. Write your Roll No. on the top immediately on receipt of this question paper.
2. Attempt five questions in all.
3. Question No. 1 is compulsory.
4. Give illustrations and examples wherever required.
5. (Subparts of the questions should be attempted together).

Q 1. (a) Define the following (any three):

(3 × 2 = 6)

- i. RAPD
- ii. YAC
- iii. FISH
- iv. STS

(b) State whether TRUE or FALSE and justify (any three)

(3 × 2 = 6)

- i. Sex-influenced traits are controlled by the genes present on allosomes.
- ii. Consanguinity increases the risk of dominant disorders.
- iii. Mutations do not affect Hardy–Weinberg equilibrium.
- iv. UPD involves inheritance of diploid chromosome from one parent only.

(c) Comment on any two:

(2 × 3 = 6)

- i. Effects of consanguinity.
- ii. Linkage disequilibrium
- iii. Effects of genetic polymorphism in drug metabolism gene

Q 2. (a) Describe the history of human genetics, highlighting contributions of Francis Galton and Archibald Garrod. (6)

(b) Why is prenatal diagnosis important? Elaborate any one invasive method of prenatal diagnosis. (6)

(c) How is retinoblastoma different from color blindness? (6)

P.T.O.

Q 3. (a) How is the hierarchical approach of sequencing human genome different from the shotgun sequencing? (9)

(b) Discuss the use of PCR in screening SNPs and microsatellite markers. (9)

Q 4. Explain in detail (9 x 2 = 18)

- i. Vectors used for sequencing the human genome
- ii. DNA fingerprinting

Q 5. (a) How will the sequencing gels of the following fragment look like in Maxim-Gilbert and Sanger's sequencing method? In what ways are these two sequencing methods different from one another? (9)

5' GAATTCAGGTACATTC 3'

(b) How are autosomal dominant and autosomal recessive traits different from one another? Explain with the help of representative pedigrees. (9)

Q 6. (a) A population of wild cats can be either black or white; the black allele (B) has complete dominance over the white allele (b). Given a population of 1,000 wild cats, 840 black and 160 white, determine: allele frequency, the frequency of individuals per genotype, and number of individuals per genotype. (B) The next generation of cats has a total population of 800 cats, 680 black and 120 white. Is the population in Hardy-Weinberg Equilibrium? (9)

(b) Plasmid pRIT451 was cut with SmaI, BglII, and AvaI. From the data below, determine the restriction map of the plasmid. (9)

SmaI	5.9	4.3		
BglII	8.2	2.0		
AvaI	5.3	4.9		
SmaI + BglII	5.4	2.8	1.5	0.5
SmaI + AvaI	3.3	2.6	2.3	2.0
AvaI + BglII	5.3	2.1	2.0	0.8