B.TECH/BT/7TH SEM/BIOT 4122/2025

HUMAN GENOMICS
(BIOT 4122)

Time Allotted : 2½ hrs **Full Marks : 60**

Figures out of the right margin indicate full marks.

Candidates are required to answer Group A and any 4 (four) from Group B to E, taking one from each group.

Candidates are required to give answer in their own words as far as practicable.

Group – A

1. Answer any twelve: **12 × 1 = 12**

Choose the correct alternative for the following

(i) How many types of deoxynucleoside triphosphates are used in Sanger sequencing?
(a) 1 (b) 2 (c) 3 (d) 4

(ii) Sequence of which of the following cannot be determined using the Maxam Gilbert method?
(a) Bacteria (b) Plants
(c) Bacteriophage T7 (d) Plasmid

(iii) Which of the following are the sites for gene variations?
(a) drug target protein (b) drug transport protein
(c) drug metabolize enzyme (d) all of these

(iv) Exon skipping is…
(a) silent mutations (b) RNA processing mutations
(c) nonsense mutations (d) regulatory mutation

(v) Genetic polymorphisms account for differences in metabolism, including
(a) Poor metabolizers (PMs) are those
(b) who lack a working enzyme
(c) Intermediate metabolizers (IMs) who have one working, wild-type allele and one mutant allele
(d) Extensive metabolizers (EMs), with two normally functioning alleles

(vi) At how many locations, SNPs are present in human beings?
(a) 1.4 million (b) 1.4 billion
(c) 3×10^9 bp (d) 3×10^4 bp

(vii) Which of the following is typically found as part of the prokaryotic genome?
(a) Chloroplast DNA (b) Linear chromosomes
(c) Plasmids (d) Mitochondrial DNA

(viii) A ____ is a group of genes or DNA sequences that are inherited together on the same chromosome.
(a) Haplotype (b) Genotype
(c) Heteroplast (d) Phenotype

(ix) A way to discern lateral gene transfer is through phylogenetic analysis, referred to as an 'among-genome' approach, which can be used to discover __________
(a) recent lateral gene transfer events but almost negligible ancient events
(b) recent lateral gene transfer events
(c) ancient lateral gene transfer events
(d) both recent and ancient lateral gene transfer events

(x) Which of the following is untrue regarding EST Index Construction?
(a) The goal of the EST databases is to organize and consolidate the largely redundant EST data
(b) The process includes a preprocessing step that removes masks repeats
(c) There is no screening of vector contaminants
(d) The goal of the EST databases is to improve the quality of the sequence information so the data can be used to extract full-length cDNAs

Fill in the blanks with the correct word

(xi) XXY karyotype is seen in persons with _________ syndrome.

(xii) The transfer of naked DNA from one cell to another is referred to as ________.

(xiii) The number of Barr Bodies present in a XXXYY individual is ________.

(xiv) In some gene families the genes are clustered in defined chromosomal regions as a result of ______ gene duplication.

(xv) ________% of human genome codes for proteins.

Group - B

2. (a) Name a process of identifying gene location. *[(CO1) (Knowledge/LOCQ)]*
(b) Determine the three forms of this process and cite their characteristics with a diagram showing the relationship among those processes. *[(CO1) (Determine/IOCQ)]*
(c) Describe the steps for processing data for construction of the UniGene database. *[(CO2)(Describe/LOCQ)]]*

1 + (3 + 3 + 3) + 2 = 12

3. (a) Discuss the major points of comparison of human genome with other sequenced genomes. *[(CO1) (Knowledge/LOCQ)]*
(b) We now know that less that 2% of the genome codes for proteins. What is the utility of the rest of the genome? Analyse with reasons. *[(CO1) (Determine/IOCQ)]*
(c) Describe the steps for processing data for construction of the UniGene database. *[(CO2)(Describe/LOCQ)]]*

4 + 4 + 4 = 12

Group - C

4. (a) Explain the basis of using 200 to 300 nucleotide-long sequences gathered from cDNA clones for transcriptome analysis. *[(CO3) (Analyse/IOCQ)]*
   (b) Describe the relationship of various mapping techniques with diagrams. *[(CO3) (Remember/LOCQ)]*
   (c) Examine the role the above method as a gene mapping tool. *[(CO1,4)(Examine/HOCQ)]*

4 + 4 + 4 = 12

5. (a) Describe the experimental procedure of SAGE and mention the technical difficulties of SAGE. *[(CO2) (Remember/LOCQ)]*
   (b) 'In cancer studies SAGE plays a major role'. Justify this role with suitable example. *[(CO2) (Analyze/IOCQ)]*

(5 + 2) + 5 = 12

Group - D

6. (a) Mention the salient features of the mitochondrial genome. *[(CO3) (Remember/LOCQ)]*
   (b) Comment on the functions of snRNA genes and snoRNA genes. *[(CO3) (Remember/LOCQ)]*
   (c) Briefly comment on 'partially overlapping genes' and 'genes within genes' with suitable examples. *[(CO3)(Understand/LOCQ)]*

4 + 4 + 4 = 12

7. (a) What do you mean by genes within genes? Give example. *[(CO4) (Remember/LOCQ)]*
   (b) Explain with examples what do you mean by a gene family and gene superfamily. *[(CO4) (Analyze/IOCQ)]*
   (c) The snRNA and snoRNA genes provide important information about DNA repeats. Criticize the statement with reasons. *[(CO4)(Criticize/HOCQ)]*

4 + 4 + 4 = 12

Group - E

8. (a) Analyse the role of the candidate genes reported by genomics research in a polygenic disorder. *[(CO5)(Analyse/IOCQ)]*
   (b) Give a critical assessment on the innovations in pharmacogenomics and its effects on human healthcare. *[(CO5) (Criticise/HOCQ)]*

(3 + 3) + 6 = 12

9. (a) What are Haplotypes? How are they helpful in association studies for locating disease genes? *[(CO4) (Remember/LOCQ)]*
   (b) Analyze one dye-based method for detection of SNP. *[(CO4) (Analyze/IOCQ)]*
   (c) How can you estimate the coefficient of linkage disequilibrium between two alleles? *[(CO6) (Calculate/IOCQ)]*

4 + 4 + 4 = 12

Cognition Level	LOCQ	IOCQ	HOCQ
Percentage distribution	44	42	14