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) Variation between individuals due to single base changes is called as
(a) ESTs (b) Contigs (c) SNPs (d) Transversion

(ii) HGP stands for _____.
(a) Human Gene Project (b) Homogeneous Genome Project
(c) Human Genome Project (d) Human Genetic Project

(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) 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.

(v) Complete genomic sequence of which plant has been completed
(a) *Arabidopsis thaliana* (b) *Tritium vulgare*
(c) *Cucurbita pepo* (d) *Pisum sativum*.

(vi) 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.

(vii) 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.

(viii) Name the sequences which are present in more than one copy in a haploid genome?
(a) Nonrepetitive DNA (b) Highly repetitive DNA
(c) Repetitive DNA (d) Minisatellite.

(ix) Which of the following are examples of genetic polymorphisms?
(a) Glutathione S-transferase (b) Dihydropyrimidine dehydrogenase
(c) UDP-glucuronosyl transferase (d) All of these.

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

Fill in the blanks with the correct word

(xi) _________ was a suitable vector for the process of cloning in Human Genome Project.

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

(xiii) _____ gene is also known as the 'Guardian of the genome'.

(xiv) An interaction between non-allelic genes in which an allele at one locus prevents expression of an allele at another locus, but not vice versa, is called ____________.

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

Group - B

2. (a) Illustrate the basic principle of Sanger's dideoxy method of sequencing.
[(CO1,2)(Remember/LOCQ)]
(b) Describe the technique used in Whole Genome Shotgun sequencing.
[(CO2)(Describe/LOCQ)]
(c) What are the advantages of 454 sequencing over conventional methods?
[(CO2)(Understand/LOCQ)]
4 + 4 + 4 =12

3. (a) Analyze how clone, contig and supercontig perform genome assembly.
[(CO1,2)(Analyze/IOCQ)]
(b) Mention the utility of the following software and justify their role in genome assembly- Phred and Phrap. *[(CO2)(Understand/LOCQ)]*
(2 + 2 + 2) + (3 + 3) = 12

Group - C

4. (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

5. (a) Genome economy–synthesizing more proteins from fewer genes. Discuss the mechanisms that are responsible for genome economy by citing suitable examples. *[(CO2)(Discuss/IOCQ)]*

(b) "Gene function description in genome annotation is often ambiguous and imprecise as it uses natural language". Using one suitable example evaluate how gene ontology project solves this problem. *[(CO2)(Evaluate/HOCQ)]*

(4 + 2) + (3 + 3) = 12

Group - D

6. (a) The Nobel Prize in Physiology and Medicine (2022) was awarded to Svante Pääbo for his discoveries in the area of Paleogenomics. Give your critical assessment on the significance of Paleogenomics in tracing human evolution. *[(CO3)(Criticize/HOCQ)]*

(b) Analyze the methodological challenges in the analysis of archaic DNA. *[(CO3)(Analyze/IOCQ)]*

(c) Discuss the relationship of the Denisovan genome with modern human and Neanderthal genome. *[(CO3)(Understand/IOCQ)]*

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) Give a critical appreciation of the Genome wide association studies (GWAS) in the identification of genetic variants among populations. *[(CO4)(Criticize/HOCQ)]*

(b) Write a brief note on any one flourogenic dye-based genotyping method. *[(CO4)(Remember/LOCQ)]*

(c) Illustrate the 'Invader Assay' in the detection of SNP. *[(CO4)(Understand/LOCQ)]*

4 + 4 + 4 = 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	42.70	38.54	18.75