

**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) Commonly used vectors for human genome sequencing are
(a) T-DNA (b) BAC and YAC
(c) Expression vectors (d) T/A cloning vectors.
- (ii) Mark the INCORRECT statement about minisatellites
(a) Tandemly repeated DNA (b) Form clusters up to 200 kb length
(c) Shorter clusters (d) Found in the centromere region.
- (iii) The phenomenon which shows the lack of correlation in genome size and complexity is
(a) Histogram (b) Karyogram
(c) Dendogram (d) C-value paradox.
- (iv) Complete genomic sequence of which plant has been completed
(a) Arabidopsis thaliana (b) Tritium vulgare
(c) Cucurbita pepo (d) Pisum sativum.
- (v) Which is the basis of genetic mapping of the human genome as well as DNA fingerprinting?
(a) Polymorphism in the DNA sequence
(b) Single nucleotide polymorphism
(c) Polymorphism in the hnRNA sequence
(d) Polymorphism in the RNA sequence.
- (vi) Which of the following is UNTRUE regarding expressed sequence tags (ESTs)?
(a) One of the high throughput approaches to genome-wide profiling of gene expression is sequencing ESTs
(b) They are short sequences obtained from cDNA clones
(c) They serve as short identifiers of full-length genes
(d) They are typically in the range of 800 to 900 nucleotides in length.

- (vii) Variation between individuals due to single base changes is called as
 (a) ESTs (b) Contigs (c) SNPs (d) Transversion.
- (viii) Conserved gene order can be termed as _____
 (a) Ortholog (b) Synteny
 (c) Paralog (d) Microarray.
- (ix) STS stands for
 (a) Simple Tagged Sequence (b) Sequence Tagged Sites
 (c) Simple Tagged Service (d) None of these.
- (x) Cytologic maps can be considered to be of ____ resolution and hence somewhat _____ physical maps.
 (a) very high, inaccurate (b) very low, accurate
 (c) very high, accurate (d) very low, inaccurate

Fill in the blanks with the correct word

- (xi) Chain termination method was discovered by _____.
- (xii) _____ DNA is present at the telomere region of a chromosome.
- (xiii) The full form of QTL is _____.
- (xiv) _____ genomics includes a comparison of gene number, gene content, and gene location in both prokaryotic and eukaryotic groups of organisms.
- (xv) The largest known human gene is _____.

Group - B

2. (a) Analyze the role of BAC or YAC in mapping techniques. [[CO1](Analyze/IOCQ)]
 (b) The hybrid sequencing technique is an amalgamation of the hierarchical and whole genome sequencing approaches – Justify the statement with reasons. [[CO1](Justify/HOCQ)]
 (c) Draw a comparative study between the two sequencing approaches followed by NIH and Celera Genomics during the HGP. [[CO1](Compare/IOCQ)]
4 + 4 + 4 = 12
3. (a) State the 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

Group - C

4. (a) Analyse the role of comparison of whole genomes in Comparative Genomics. [[CO2](Analyse/IOCQ)]
 (b) ‘Minimal set of genes are required for maintaining the free living cellular organism’ - explain the statement. [[CO2](Remember/LOCQ)]

- (c) Explain with the help of suitable example how syntenic relationships provide important clue to functional relationships of genes of interest. [[CO1,2](Examine/HOCQ)]
4 + 4 + 4 = 12
5. (a) Explain the basis of using 200 to 300 nucleotides long sequences gathered from cDNA clones can be used for transcriptome analysis. [[CO3](Analyse/IOCQ)]
- (b) Diagrammatically describe the relationship of various mapping techniques. [[CO3](Remember/LOCQ)]
- (c) Examine the role the above method as a gene mapping tool. [[CO1,4](Examine/HOCQ)]
4 + 4 + 4 = 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) Give a comparative account of functionally similar, related and identical genes. [[CO3](Compare/IOCQ)]
- (b) Mention the salient features of the mitochondrial genome. [[CO3](Remember/LOCQ)]
- (c) Comment on the functions of snRNA genes and snoRNA genes. [[CO3](Remember/LOCQ)]
4 + 4 + 4 = 12

Group - E

8. (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](Analyse/IOCQ)]
- (c) Evaluate how you can quantify the relationship between two alleles by linkage disequilibrium (LD). [[CO4](Evaluate/HOCQ)]
4 + 4 + 4 = 12
9. (a) Analyse the role of the candidate genes reported by genomics research in a polygenic disorder. [[CO5](Analyse/IOCQ)]
- (b) Discuss the impact of pharmacogenomics in drug discovery and disease diagnosis [[CO5](Comprehension/LOCQ)]
(3 + 3) + 6 = 12

Cognition Level	LOCQ	IOCQ	HOCQ
Percentage distribution	30.20	48.96	20.84

Course Outcome (CO):

At the end of this course students will be able to:

1. Develop a concept of the different genome mapping techniques and the genome assembly methods.
2. Understand the usage of functional genomics tools, different methods of gene transfer and applications of comparative genomics.
3. Understand the background of the Human Genome Project along with its findings on genome anatomy, gene family, gene diversity and gene markers.
4. Analyze the haplotypes and SNPs by various quantitative techniques.
5. Interpret the findings of Human Genome Project in the domain of pharmacogenomics and polygenic disorders.

**LOCQ: Lower Order Cognitive Question; IOCQ: Intermediate Order Cognitive Question; HOCQ: Higher Order Cognitive Question.*