HUMAN GENOMICS
(BIOT 4122)

Time Allotted : 3 hrs **Full Marks : 70**

Figures out of the right margin indicate full marks.

Candidates are required to answer Group A and any 5 (five) from Group B to E, taking at least one from each group.

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

Group – A
(Multiple Choice Type Questions)

1. Choose the correct alternative for the following: **10 × 1 = 10**

(i) The first completed genome sequencing project was of
(a) E. Coli (b) Haemophilus influenzae
(c) Mus musculus (d) Drosophila melanogaster.

(ii) The best estimate for the number of human genes is _____.
(a) 100,000 (b) 50,000 (c) 20,000 (d) 10,000

(iii) Which of the following is untrue about Lateral gene transfer?
(a) It is also known as vertical gene transfer
(b) There is exchange of genetic materials between species
(c) It mainly occurs among prokaryotic organisms when foreign genes are acquired through mechanisms
(d) It is one of the examples is transformation.

(iv) The private company involved in human genome sequencing in parallel with NIH was
(a) Roche (b) Celera (c) Gilead (d) Genentech.

(v) According to HGP, genetic similarity between all humans is
(a) 90% (b) 95% (c) 99.9% (d) 99.5%.

(vi) QTL is related to
(a) Quantifying alleles in complex disorders
(b) Quantifying amount of DNA
(c) Quantifying allele frequencies in a population
(d) Quantifying single gene disorders.

(vii) The network of interactions engaged in by protein at cellular level is described in _____.
(a) Phenotypic function (b) Cellular function
(c) Molecular function (d) Structural genomics

(viii) Genes of different species but possessing a clear sequence and functional relationship to each other are _____.
(a) Ortholog (b) Synteny (c) Paralog (d) Microarray

(ix) Which of the following is incorrect regarding gene annotation?
(a) The gene annotation of the human genome employs a combination of theoretical prediction and experimental verification
(b) Gene structures are first predicted by *ab initio* exon prediction programs
(c) The predicted genes are compared with experimentally determined cDNA and EST sequences
(d) The pairwise alignment programs are not involved.

(x) A comprehensive database for the study of human genetics and molecular biology is
(a) PDB (b) STAG (c) OMIM (d) PSD.

Group- B

2. (a) Describe the steps of clone-by-clone shotgun sequencing. [(CO1)(Remember/LOCQ)]
(b) Analyze the process of Pyrosequencing with a suitable diagram. [(CO1)(Analyze/IOCQ)]
(c) 'emPCR is a big step towards high-throughput genome sequencing'. Justify the statement with reasons. [(CO1)(Justify/HOCQ)]
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 softwares and justify their role in genome assembly- Phred and Phrap. [(CO2)(Understand/LOCQ)]
6 + (3 + 3) = 12

Group - C

4. (a) Genome economy –synthesize 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

5. (a) "Comparison of whole genomes generate huge knowledge -this can be potentially useful in future metabolic pathway engineering"- analyze this statement. [(CO2)(Analyze/IOCQ)]
(b) State the system by which the lateral gene transfer occurs. [(CO2)(Analyse/LOCQ)]
i. Assess how this event has a relationship with its recent occurrences. [(CO2)(Evaluate /HOCQ)]
ii. Discuss how gene order plays an impact in comparative genomics. [(CO2)(Discuss/LOCQ)]
3 + 2 + 3 + 4 = 12

Group - D

6. (a) Why was building cDNA libraries an important step in genome sequencing? [(CO3)(Understand/LOCQ)]
   (b) Analyze the significance of SNPs on medicine and therapeutics. [(CO4)(Analyze/IOCQ)]
   (c) Comment on the ethical, legal and social implications of HGP. [(CO3)(Analyze/IOCQ)]
   4 + 4 + 4 = 12

7. (a) What do you mean by nonprocssed pseudogenes? Give example. [(CO4)(Remember/LOCQ)]
   (b) Explain with examples what do you mean by a gene family and gene superfamily. [(CO4)(Analyze/IOCQ)]
   (c) Give a comparative assessment between satellite, minisatellite and microsatellite DNA. [(CO4)(Criticize/HOCQ)]
   4 + 4 + 4 = 12

Group - E

8. (a) Discuss the technique of dCAPS. [(CO4)(Remember/LOCQ)]
   (b) Comment on the single base extension method for detection of SNP. [(CO4)(Analyze/IOCQ)]
   (c) Write a brief note on TaqMan assay. [(CO4)(Remember/LOCQ)]
   4 + 4 + 4 = 12

9. (a) State a disease where human disease genes have been identified with the help of animal models. Explain how it is done. [(CO1)(Remember/LOCQ)]
   (b) For identification of disease genes, these genes from candidate region must be prioritized for mutation testing- discuss how this is done citing suitable example. [(CO2)(Understand/LOCQ)]
   (1 + 3) + (4 + 4) = 12

Cognition Level	LOCQ	IOCQ	HOCQ
Percentage distribution	45.33	36.45	17.7

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.