

**HUMAN GENOMICS
(BIOT 4165)**

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 ____ resolution genome map is the genomic DNA sequence that can be considered as a type of ____ map describing a genome at the single base-pair level.
 (a) highest, physical (b) lowest, physical
 (c) highest, cytological (d) lowest, cytological
- (ii) The shotgun approach ____ sequences clones from ____ of cloned DNA.
 (a) randomly, one end (b) randomly, both ends
 (c) specifically, both ends (d) specifically, one end
- (iii) The largest gene in the human genome is
 (a) Titin (b) Dystrophin
 (c) Insulin (d) Phosphofructokinase.
- (iv) A cDNA library contains clones representing which of the following?
 (a) mRNA (b) Genomics DNA
 (c) Repeated sequences (d) Introns.
- (v) QTL stands for
 (a) Qualitative Trait Loci (b) Quantitative Trait Loci
 (c) Qualitative Trait Location (d) Quantitative Trait Location.
- (vi) Variation between individuals due to single base change is called
 (a) EST (b) SNP (c) contig (d) transversion.
- (vii) Which of the following repeated sequences includes an open reading frame for reverse transcriptase?
 (a) SINE (b) LINE (c) Transposon (d) VNTR.

- (viii) Which of the following is untrue about cytologic maps?
 (a) They cannot be directly observed under microscope
 (b) They refer to banding patterns
 (c) They can be viewed on stained chromosomes
 (d) They can be directly observed under microscope.
- (ix) Which of the given statement is incorrect?
 (a) The predicted set of proteins for the genome is referred to as the proteome
 (b) The amino acid sequence of proteins encoded by the predicted genes is used as a query of the protein sequence databases in a database similarity search
 (c) A match of a predicted protein sequence to one or more database sequences serves only to identify the gene function but it doesn't validate the gene prediction
 (d) The genome sequence is annotated with the information on gene content and predicted structure, gene location, and functional predictions.
- (x) 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.

Group – B

2. (a) How can you prepare a cDNA library?
 (b) What is STS? Mention its utility in physical mapping.
 (c) Describe the technique of clone-by-clone DNA sequencing with a diagram.
3 + (2 + 2) + 5 = 12
3. (a) Illustrate the basic principle of Sanger's dideoxy method of sequencing.
 (b) Describe the technique used in Whole Genome Shotgun sequencing.
 (c) What are the advantages of 454 sequencing over conventional methods?
4 + 4 + 4 = 12

Group – C

4. (a) "Comparison of whole genomes generate huge knowledge - this can be potentially useful in future metabolic pathway engineering" - explain this statement.

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Group – C

4. (a) "Comparison of whole genomes generate huge knowledge - this can be potentially useful in future metabolic pathway engineering" - explain this statement.

- (b) In which system the lateral gene transfer occurs?
- (c) Mention how this event has a relationship with its recent occurrences.
- (d) Mention how gene order plays an impact in comparative genomics.

3 + 2 + 3 + 4 = 12

5. (a) State a disease where human disease genes have been identified with the help of animal models and explain how it is done.
- (b) For identification of disease genes, these genes from candidate region must be prioritised for mutation testing - discuss how this is done citing suitable examples.

(1 + 3) + 8 = 12

Group - D

6. (a) Elaborate on the ELSI aspect of the Human Genome Project.
- (b) On the basis of the findings of HGP, how does human genome differ from lower organisms?
- (c) Comment on the gene density in the human genome.

4 + 4 + 4 = 12

7. (a) What do you mean by genes within genes? Give an example.
- (b) Write a brief note on the salient features of mitochondrial genome.
- (c) What is a classical gene family? How does it differ from a gene superfamily?

(3 + 1) + 4 + (2 + 2) = 12

Group - E

8. (a) Discuss the importance of SNPs in association studies and disease genomics.
- (b) What is a haplotype? What are the benefits of HapMap project in genomics research?
- (c) What is QTL mapping?

5 + (1 + 4) + 2 = 12

9. (a) What do you mean by personalized medicine?
- (b) Describe the principle behind linkage disequilibrium.
- (c) Discuss the importance of association studies in genomics research.

4 + 4 + 4 = 12