

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 phenomenon which shows the lack of correlation in genome size and genetic complexity is called
(a) Histogram (b) Karyogram
(c) Dendogram (d) C-value paradox
- (iii) In hierarchical genome sequencing approach, based on the results of _____ mapping, _____ of the BAC clones on a chromosome can be determined.
(a) physical, the locations and orders (b) physical, only the locations
(c) cytological, only the locations (d) physical, only the orders
- (iv) A cDNA library contains clones representing which of the following?
(a) mRNA (b) Genomic DNA
(c) Intron (d) microRNA
- (v) Which of the following repeated sequences includes an open reading frame for reverse transcriptase?
(a) LINE (b) SINE
(c) Segmental duplication (d) DNA transposon
- (vi) Which of the following is not a gene expression database?
(a) Flyview (b) Genbank
(c) Bodymap (d) None of these
- (vii) Short sub-sequence of a cDNA sequence is
(a) Expressed sequence tag (b) Sequence tagged site
(c) Contig (d) YAC

- (viii) Which of the following is untrue about comparative genomics?
(a) It is comparison of whole genomes from different organisms.
(b) It includes comparison of gene number, gene location, and gene content from these genomes.
(c) It provides insights into the mechanism of genome evolution and gene transfer among genomes.
(d) It doesn't help to reveal the extent of conservation among genomes.
- (ix) In which of the following models might a compensatory mechanism mask the effect of a genetic manipulation?
(a) Knockouts only (b) Knockins only
(c) Knockouts and Knockins (d) All model
- (x) Which of these projects would be best suited for Next Generation Sequencing?
(a) To determine if a tumour sample contains a common missense mutation
(b) To find the transcriptome of a tumour sample
(c) To genotype ten genomic DNA samples for a known SNP
(d) All of the above.

Group- B

2. (a) Comment about the challenges in human genome sequencing. [(CO1) (Understand/LOCQ)]
(b) Analyze the process of whole genome shotgun sequencing. [(CO1) (Analyze/IOCQ)]
(c) The 454 sequencing has revolutionized DNA sequencing – Justify the statement with reasons. [(CO1)(Justify/HOCQ)]
4 + 4 + 4 = 12
3. (a) The high resolution physical mapping involves two approaches - describe them with suitable diagrams. [(CO1) (Explain/IOCQ)]
(b) Write the characteristics of chromosomal map and cDNA map along with their importance. [(CO1) (Remember/LOCQ)]
(3 + 3) + (3 + 3) = 12

Group - C

4. (a) Design one high throughput sequence based approach used in functional genomics where the sample size is 200-400bp and cite its characteristics. [(CO2) (Design/HOCQ)]
(b) This approach plays an immense role in organising redundant informative sequence information. Analyse how it is done. [(CO2) (Analyse/HOCQ)]
(c) Analyze how the NCBI informative cluster database is constructed following this approach. [(CO2) (Analyse/IOCQ)]
(1 + 3) + 4 + 4 = 12

5. (a) Describe the steps of the different levels of genome sequence assembly along with its constraints. [(CO2) (Explain/IOCQ)]
- (b) "The field of genomics comprises not a single area rather encompasses analysis of gene information and analysis by virtue of tools". Mention the areas that fall under this category. Cite two characteristics and correlation for those areas. [(CO2) (Correlate/IOCQ)]
- 4 + (4 + 4) = 12**

Group - D

6. (a) The HGP showed that less than 2% of the human genome codes for functional proteins – Criticize the statement with reasons. [(CO3) (Criticize/HOCQ)]
- (b) Analyze the significance of microbial genomics in human welfare and research. [(CO3) (Analyze/IOCQ)]
- (c) Discuss how mtDNA and Y-chromosomal DNA markers help in tracing human evolution. [(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) What were the main objectives of HapMap project? [(CO4) (Remember/LOCQ)]
- (b) Analyze one dye-based method for detection of SNP. [(CO4) (Analyze/IOCQ)]
- (c) Discuss the concept of QTL in quantitative genomics approach. [(CO4)(Analyze/IOCQ)]
- 4 + 4 + 4 = 12**
9. (a) "It is been observed that genes affect our reaction to drugs". Criticize the statement with suitable example. [(CO5)(Evaluate/HOCQ)]
- (b) Elucidate the clinical implications of Pharmacogenomics with suitable example. [(CO5)(Analyze/IOCQ)]
- (c) Cite two FDA-approved diagnostic tests commercially available for commonly prescribed pharmacologic therapies. [(CO5) Remember/LOCQ]
- 4 + 4 + 4 = 12**

Cognition Level	LOCQ	IOCQ	HOCQ
Percentage distribution	27%	52%	21%

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

Department & Section	Submission Link
BT	https://classroom.google.com/c/NDIwMzczMTYxNjly/a/NDY0MTU5MzlyNjUx/details