SARDAR PATEL UNIVERSITY

[1013]

B. Sc. (Genetics) – Fifth Semester Examination (CBCS)
Tuesday, 29th December 2020
2:00 p.m. to 4:00 p.m.
US05CGEN24 : Human Genetics and Bioinformatics

Total Marks: 70

Note: (1) Figures to the right indicate marks.

       (2) Draw a neat and labeled diagram, wherever necessary.

Q. 1 Choose the most appropriate answer from the four alternatives given: [10]

i. _____is not an example of transposon?

    (a) AFLP   (b) SINEs   (c) LINEs   (d) LTRs

ii. Which one of the following describes a contig?

    (a) Library of overlapping clones   (b) A complete mRNA library

    (c) An ordered genomic library   (d) None of these

iii. _____has the tandem repeat units with 2 – 5 base pair length.

    (a) Minisatellites   (b) SINEs   (c) LINEs   (d) Microsatellites

iv. Who invented genetic mapping?

    (a) Gregor Mendel   (b) Alfred Sturtevant   (c) C V Raman   (d) Thomas Morgan

v. Genetic defect in blood clotting factor VIII gene causes which disease?

    (a) Albinism   (b) Hemophilia 'a'   (c) Hemophilia 'b'   (d) Hemophilia 'c'

vi. Child with phenylketonuria fail to produce an enzyme _____.

    (a) DOPA   (b) Phenylalanine Hydroxylase   (c) Tyrosinase   (d) Tyrosine decarboxylase

vii. Which of the following are invasive technique?

    (a) Amniocentesis   (b) Chorionic villus sampling   (c) Both 'a' and 'b'   (d) $FeCl_3$ test

viii. Triple marker test exclude_____.

    (a) AFP   (b) Estriol   (c) Inhibin A   (d) Human chorionic gonadotrophin

ix. An ORF with 900 nucleotides can code maximum _____numbers of amino acids.

    (a) 100   (b) 200   (c) 300   (d) 900

x. The most common forms of secondary structure in proteins are _____.

    (a) α-helix   (b) β-pleated sheet   (c) α-helix and β-pleated sheet   (d) Beads on a string

Q.2 Fill in the blanks or Write True / False as applicable.   [08]

i. _____is a unit for physical map.

ii. *In situ* hybridization is used to locate specific gene/s on chromosome.   **(True / False)**

iii. Mode of inheritance for galactosemia is_____.

iv. Parkinson's disease is an example of chromosomal disorder.   **(True / False)**   *(P.T.O.)*

v. SAGE technique was developed by Victor Velculescu. **(True / False)**

vi. Low level of MSAFP indicates_____syndrome.

vii. Linear sequence of amino acids in a peptide or protein gives rise to_____structure of protein.

viii. Phylogeny is the study of relationships among different groups of organisms and their evolutionary development. **(True / False)**

Q.3 Attempt any TEN from the following Short Answer Questions (10 out of 12): [20]

i. What are gene families?

ii. Explain restriction map in short.

iii. What are coding sequences?

iv. Enlist any 4 examples of monogenic disorders.

v. What is Obesity?

vi. Enlist 3 major types of Diabetes mellitus.

vii. Write full form of MSAFP. Write its importance.

viii. Differentiate between diagnostic and predictive tests.

ix. Write importance of prenatal screening.

x. Why promoter region is important?

xi. Enlist methods for the prediction of secondary structure of proteins.

xii Write main features of open reading frame.

Q.4 Answer any FOUR from the following Long Answer Questions (4 out of 8): [32]

(a) Give an overview and significance of human genome project.

(b) Discuss preparation and applications of BAC libraries in human genome mapping.

(c) Write short notes on: **(i)** Cystic fibrosis **(ii)** Alzheimer disease.

(d) Describe Thalassemia and Klinefelter syndrome.

(e) Write short notes on: **(i)** Guthrie bacterial inhibition assay **(ii)** QUAD test.

(f) Explain: **(i)** Fetal karyotyping **(ii)** Context of prenatal diagnosis.

(g) Enlist and explain any 2 methods for the preparation of phylogenetic tree.

(h) Describe prediction of tertiary structure of proteins using homology modeling.

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