

(231)

SARDAR PATEL UNIVERSITY**M. Sc. (Genetics) – Third Semester Examination (CBCS)****Saturday, 1st December 2012****2:30 p.m. to 5:30 p.m.****PS03CGEN02: Human Molecular Genetics****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: [08]**I. Huntington's chorea is characterized by _____**

- (a) Jerky uncontrollable movement (b) CAG repeat expansion
(c) Poly Q tract (d) All of the above

II. Lisch nodule is typical symptom found in a patient with _____

- (a) Neurofibromatosis (b) Cystic fibrosis (c) Hemophilia A (d) Both (a) & (b)

III. During phase - I of drug metabolism which group is added by cytochrome P450 enzyme

- (a) Acetyl (b) Ester (c) Hydroxyl (d) Methyl

IV. 6- mercaptopurine and 6- thioguanine are detoxified by an enzyme _____

- (a) N- acetyl transferase (b) G6PDH (c) Choline esterase (d) TPMT

V. Linkage mapping can determine the distance between which of the following pairs of DNA sequences?

- (a) Two known genes (b) AFLPs and RFLPs (c) Two AFLPs (d) All of the above

VI. Which one of this describes a contig?

- (a) Library of overlapping clones (b) A complete mRNA library
(c) An ordered genomic library (d) None of these

VII. The incidence of albinism can be artificially increased in fish by exposing eggs to _____

- (a) Arsenic (b) Cadmium (c) Copper and mercury (d) All of the above

VIII. Match the following and choose correct answer from the codes given below:

- | | |
|-----------------------|--|
| A. GSD in Live stocks | 1. Accumulation of branched chain aminoacids |
| B. MSUD | 2. Loss of skin, hair and eyes pigmentation |
| C. Alkaptonuria | 3. Intoxication with castanospermine |
| D. PKU | 4. Homogentisic aciduria |

- | | A | B | C | D |
|-----|---|---|---|---|
| (a) | 1 | 2 | 3 | 4 |
| (b) | 2 | 4 | 1 | 3 |
| (c) | 3 | 1 | 4 | 2 |
| (d) | 4 | 3 | 2 | 1 |

P.T.O.

- Q.2 Answer any SEVEN from the following:** [14]
- Define positional cloning. Enlist two applications of positional cloning.
 - Differentiate between genetic and physical mapping.
 - Enlist functions of Htt protein.
 - Differentiate between WT gene and RB gene.
 - Write four important mutations in PAH gene.
 - Write full names of: BCKDHA, BCKDHB, DBT and DLD
 - Differentiate between pharmacodynamics and pharmacokinetics.
 - Define terms: Ecogenetics and pharmacogenomics
 - Classify hemophilia.

- Q.3(a)** Enlist various genetic markers. Discuss their importance in gene mapping. [6]
- (b)** Describe pulse field gel electrophoresis. [6]

OR

- (b)** 1. Explain synteny of genes. [3]
2. Write brief note on chromosome walking. [3]

- Q.4(a)** "Obesity is a polygenic disorder" Justify the statement. [6]
- (b)** Discuss genetic aspects of IDDM and NIDDM. [6]

OR

- (b)** Write short notes on the following:
- Disease causing mutations in CFTR gene [3]
 - Genetic mutations associated with hemophilia [3]

- Q.5(a)** Identify the enzymes and their cytogenetic locations in following biochemical reactions. [6]
- What happens when all the reactions are blocked due to mutations in genes encoding these enzymes?

- Ceramide - Glc - GalNANA - GalNAc \longrightarrow Ceramide - Glc - GalNANA
- Gal - Gal - Glc - Ceramide \longrightarrow Lactosyl Ceramide
- Ceramide - Phosphocholine \longrightarrow Ceramide
- Ceramide - Gal - (S) \longrightarrow Ceramide - Gal

- (b)** Write a detail note on human mitochondrial syndromes. [6]

OR

P.T.O.

(b) Write short notes on the following:

- 1) Sanfilippo syndrome [3]
- 2) Pompe disease [3]

Q.6(a) By giving suitable example explain individual genetic variations affecting both pharmacodynamic and pharmacokinetic of a drug. [6]

(b) Discuss human genome project and its practical implications. [6]

OR

(b) Write short notes on the following:

- 1) Drug induced hemolytic anemia [3]
- 2) Vitamin K cycle [3]
