

(A)  
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**SARDAR PATEL UNIVERSITY**

**M. Sc. (Integrated Biotechnology) – Tenth Semester Examination (CBCS)**

**Tuesday, 29<sup>th</sup> March 2016**

**10:30 a.m. to 1:30 p.m.**

**PS10CIGMB1: Human 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. In positional - independent routes disease genes are identified through\_\_\_\_\_.**  
 (a) Knowing the protein products (b) The function or interaction of its product  
 (c) An animal model& DNA sequences (d) All of these
- II. The threshold for declaring linkage is an LOD score of \_\_\_\_\_.**  
 (a) Three (b) Four (c) Three or greater than three (d) Less than two
- III. Which one of the following is not a subtype of Diabetes mellitus?**  
 (a) IDDM (b) NIDDM (c) MODY (d) Diabetes insipidus
- IV. Café-au-lait is a typical characteristic feature found in a patient with\_\_\_\_\_.**  
 (a) Neurofibromatosis (b) Cystic fibrosis (c) Hemophilia A (d) Both (a) & (b)
- V. Excess of phenylalanine in the blood during PKU is detected by\_\_\_\_\_ test.**  
 (a) Guthrie (b) Ferric chloride (c) Seliwanoff's (d) DNPH
- VI. 6- mercaptopurine and 6- thioguanine are detoxified by an enzyme\_\_\_\_\_.**  
 (a) N- acetyl transferase (b) G6PDH (c) Choline esterase (d) TPMT
- VII. Antimalarial drug induced hemolytic anemia in African males due to the deficiency glucose 6 phosphate dehydrogenase is an example of variation affecting\_\_\_\_\_.**  
 (a) Pharmacodynamic response (b) Phase I metabolism  
 (c) Phase II metabolism (d) Pharmacokinetic response
- VIII. Match the following and choose correct answer from the codes given below:**
- |                               |                                      |
|-------------------------------|--------------------------------------|
| A. MPS-VII                    | 1. Heparan sulfamidase deficiency    |
| B. Sanfilippo Syndrome Type A | 2. Deficiency of iduronate sulfatase |
| C. Alkaptonuria               | 3. $\beta$ - glucuronidase           |
| D. MPS - II                   | 4. Homogentisic acid oxidase         |

- |     |   |   |   |   |
|-----|---|---|---|---|
|     | 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]**

- i. What is chromosome walking?
- ii. What are CEPH families?
- iii. Differentiate between hemophilia A and hemophilia B.
- iv. Enlist major categories of cancer causing genes.
- v. Give an overview of MSUD.
- vi. Why early diagnosis of neonatal IEM is crucial?
- vii. Write types of albinism.
- viii. Differentiate between pharmacogenetics and pharmacogenomics.
- ix. Enlist typical phase II conjugation reactions for inactivation and excretion of drugs.

**Q.3 (a) Enlist various genetic markers. Discuss any two of them with their importance in gene mapping. [6]**

**(b) What are contigs? Explain assembly of clone contigs. [6]**

**OR**

**(b) Discuss high-throughput DNA sequencing techniques. [6]**

**Q.4 (a) Enlist any 4 factors influencing on genetic susceptibility to common diseases. Describe cystic fibrosis. [6]**

**(b) Discuss genetic aspects of obesity. [6]**

**OR**

**(b) Write name of trinucleotide repeat expansion disorders involving intron and UTR region in human. Explain Huntington's disease in detail. [6]**

**Q.5 (a) Discuss human mitochondrial syndromes. [6]**

**(b) Write short notes on the following:**

1. Pompe disease [3]

2. Hurler's syndrome [3]

**OR**

**(b) Identify the enzymes catalyzing following biochemical reactions. What happens when all the reactions are blocked due to mutations in genes encoding these enzymes? [6]**

i. Gal - Gal - Glc - Ceramide  $\rightarrow$  Lactosyl Ceramide

ii. Ceramide - Phosphocholine  $\rightarrow$  Ceramide

iii. Ceramide - Gal - (S)  $\rightarrow$  Ceramide - Gal

**P.T.O.**

**Q.6(a)** With the help of suitable example, explain genetic variations affecting both pharmacokinetics and pharmacodynamics of a drug. [6]

**(b)** Discuss major social and ethical issues in medical genetics. [6]

**OR**

**(b)** Write short notes on the following:

1. Practical implications of human genome project. [3]

2. Techniques for the screening of unknown mutation. [3]

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