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SARDAR PATEL UNIVERSITY

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

Tuesday, 29th 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. β - 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 contings? 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 ⟶ Lactosyl Ceramide

ii. Ceramide - Phosphocholine ⟶ Ceramide

iii. Ceramide – Gal - (S) ⟶ 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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