Seat No.:_____ No. of Printed Pages: 2

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SARDAR PATEL UNIVERSITY

M. Sc. (Genetics) – Third Semester Examination (CBCS)

Friday, 21st October, 2016

2:00 p.m. to 5:00 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. Which of the following is not a massively parallel pyrosequencing method/platform?

(a) SOLiD (b) PAC (c) Solexa sequencing (d) Illumina genome analyzer IIX

ii. Centirays are associated with _____.

(a) EST maps (b) STS maps (c) RH maps (d) Long range maps

iii. Guthrie blood spot test is used for the neonatal screening of_____.

(a) Cystic fibrosis (b) DMD (c) Phenylketonuria (d) Hemophilia

iv. Which one of the following is a temporary diabetes_____.

(a) IDDM (b) Monogenic diabetes (c) Diabetes incipidus (d) Gestational diabetes

v. Which one of the following MPS has 4 subtypes_____.

(a) Hurlers syndrome (b) Sanfilippo syndrome

(c) Morquio syndrome (d) Sly syndrome

vi. Which one of the following is not a part of typical phase II conjugation reactions?

(a) Acetylation (b) Glucuronidation (c) Methylation (d) Hydroxylation

vii. Phase I metabolism of >90% of commonly used drugs are performed by _____.

(a) CYP1 (b) CYP2 (c) CYP3 (d) All of these

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

A. PKU	1. ATP synthase
B. MSUD	2. Iduronate sulfatase
C. Leigh's syndrome	3. Branched chain ketoacid dehydrogenase
D. MPS type II	4. Phenylalanine hydroxylase

	A	B	C	D
(a)	1	2	3	4
(b)	2	4	1	3
(c)	3	5	4	1
(d)	4	3	1	2

(PTO)

Q.2 **Answer any SEVEN from the following:** [14]

i. Differentiate between hemophilia A and hemophilia B.

ii. Mention reference families used for the construction of genetic maps.

iii. What does LOD stand for? Write its significance.

iv. What is neurofibromatosis?

v. Name any four factors influencing on genetic susceptibility to common diseases.

vi. What MELAS and NARP stands for?

vii. Why early diagnosis of IEM is crucial?

viii. Write different materials used to test the presence of genetic diseases.

ix. Differentiate between pharmacokinetics and pharmacodynamics.

Q.3(a) Enlist various types of physical maps. Explain phases in the construction of clone maps. [6]

(b) Discuss applications of pulse field gel electrophoresis. [6]

OR

(b) 1.Enlist various positional dependent strategies [3]

2. How the knowledge of protein products used to identify the disease genes? [3]

Q.4(a) What are basis for the classification of trinucleotide repeat expansion disorders. Describe Huntington disease. [6]

(b) Justify that "obesity is a polygenic and multifactorial disease". [6]

OR

(b) "Cystic fibrosis is a monogenic but multisystem disease" Justify the statement. [6]

Q.5(a) List out inherited disorders of glycogen metabolism. Describe genetic and diagnostic aspects of Pompe disease. [6]

(b) What is sphingolipodystrophy? Diagramatically explain sphingolipodystrophy associated with metabolism of ganglioside, sphingomyelin and sulfatids. [6]

OR

(b) Write short notes on the following:

1) Alkaptonuria 2) LHON [3+3]

Q.6(a) Discuss practical implications of human genome project. [6]

(b) By giving suitable example explain multiplex PCR for testing of known mutation. [6]

OR

(b) Explain SSCP and DGGE techniques for the scanning of genes for unknown mutation. [6]

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