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(A-43)

SARDAR PATEL UNIVERSITY

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

Tuesday, 21st April 2015

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]**

1. **Which of the following is a type of massively parallel pyrosequencing methods?**
   (a) SOLiD (b) PAC (c) Solexa sequencing (d) Both (a) and (c)
2. **Chromosome walking is used to_____.**
   (a) Sequence entire genome (b) Clone the gene
   (c) Sequence entire chromosomes (d) All of these
3. **Lisch nodule is typical symptom found in a patient with_____.**
   (a) Neurofibromatosis (b) Cystic fibrosis
   (c) Hemophilia A (d) Huntington's chorea
4. **MODY is an example of _____.**
   (a) IDDM (b) Monogenic diabetes (c) Diabetes incipidus (d) Gestational Diabetes
5. **Which of one the following is not a part of typical phase II conjugation reactions?**
   (a) Acetylation (b) Glucuronidation (c) Methylation (d) Hydroxylation
6. **Guthrie blood spot test is used for the neonatal screening of_____.**
   (a) Cystic fibrosis (b) DMD (c) Trinucleotide repeat expansions (d) Hemophilia
7. **Deficiency of β- glucuronidase during MPS causes_____ syndrome.**
   (a) Morquio (b) Hurler -Scheie (c) Sly (d) Maroteaux Lamy
8. **Match the following and choose correct answer from the codes given below:**

A. Farbers disease	1. α iduronidase
B. Metachromatic leukodystrophy	2. Heparan sulfatase
C. Herler syndrome	3. Arylsulfatase
D. Sanfilippo syndrome type III - A	4. Ceraminidase

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

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

1. What does LOD stand for? Write its significance.
2. Differentiate between orthologous and paralogous gene.
3. What is neurofibromatosis?
4. Name any four genes associated each with diabetes and obesity separately.
5. Differentiate between hemophilia A and hemophilia B.
6. Write example each of X linked lipidosis and mucoploysaccharidosis.
7. Write two major types of albinism in humans.
8. Write full names of TPMT and VKORC1.
9. Enlist techniques used for scanning the genes for unknown mutation.

Q.3(a) Enlist various position-independent strategies for the identification of disease genes. How the knowledge of protein products used to identify the disease genes? **[6]**

(b) Enlist various markers for gene mapping. Give an account on commonly used DNA markers for the gene mapping. **[6]**

OR

(b) 1. Explain RH mapping technique. **[3]**

2. Write applications of PFGE. **[3]**

Q.4(a) Write examples of triple repeat disorders involving exon, intron and UTR region. Explain most common CAG repeat expansion disorder. **[6]**

(b) Enlist and explain any 6 factors influencing on genetic susceptibility to common diseases. **[6]**

OR

(b) Write short notes on the following:

i. Tumor suppressor genes **[3]**

ii. Cystic fibrosis **[3]**

Q.5(a) Describe molecular and biochemical aspects in PKU and alkaptonuria. **[6]**

(b) Describe human mitochondrial syndromes. **[6]**

OR

(b) Write short notes on the following:

1) Glycogen storage disease type II **[3]**

2) Gauchar disease **[3]**

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Q.6(a) With the help of suitable example explain individual genetic variations affecting pharmacokinetics of a drug (Phase - I). **[6]**

(b) "Drug induced hemolytic anemia is due to genetic variation in pharmacodynamics response" Justify the statement. **[6]**

OR

(b) Give overview of the following:

1) Practical applications of human genome project **[3]**

2) Major ethical issues in medical genetics **[3]**

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