SEAT No.____

[A-28]

SARDAR PATEL UNIVERSITY

M. Sc. (Integrated Biotechnology) – Tenth Semester Examination (CBCS) Monday, 9th April, 2018

10:00 a.m. to 1:00 p.m.

PS10CIGMB1: Human Genetics

Total Marks: 70

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Note:	(1) Figures to the right indicate marks.(2) Draw a neat and labeled diagram, wherever necessary.	
). 1	Choose the most appropriate answer from the four alternatives given:	[08]
1.	The maximum PIC value of a biallelic marker is	
	(a) 0.5 (b) 0.375 (c) 0.53 (d) 0.053	
2.	Which of the following is a massively parallel pyrosequencing methods?	
.9	(a) SOLiD (b) PAC (c) Solexa sequencing (d) Both (a) and (c)	
3.	Centirays associated with	
	(a) EST map (b) STS maps (c) RH maps (d) Long range maps	
4.	Mutation in leptin, POMC and FTO genes increases the risk of	
٠	(a) Cystic fibrosis (b) Obesity (c) Cancer (d) Hemophilia	
5.	is an example of gender specific diabetes.	
	(a) IDDM (b) Monogenic diabetes (c) Diabetes incipidus (d) Gestational Diabetes	
6.	6 mercaptopurine and 6 thioguanine are detoxified by an enzyme	
	(a) Acetylase (b) Glucuronidase (c) TPMT (d) G6PDH	
7.	Phase I metabolism of >90% of commonly used drugs are performed by	
	(a) CYP1 (b) CYP2 (c) CYP3 (d) All of these	
8.	Match the following and choose correct answer from the codes given below:	
	A. Hemophilia 1. Nervous system	
	B. Cystic fibrosis 2. Multiple organs	
¢	C. Huntington's disease 3. Respiratory system .	
	D. Mucopolysaccharidosis 4. Hematopoetic system	
	A B C D (a) 1 2 3 4	
		•
	(b) 2 4 1 3	
	(c) 3 5 4 1	
	(d) 4 3 1 2	

Q -	2 A	ttempt ANY SEVEN from the following:	
	1.	What is candidate gene approach?	(14)
	2.	Mention reference families used for the construction of genetic maps.	
	3.	Give an over view of massively parallel sequencing techniques	
	4.	Write the symptoms of huntington's disease.	
	5.	Differentiate between hemophilia A, hemophilia B and hemophilia C.	
	6.	What is MSUD?	
	7.	Define Lipidosis and list the diseases related to it.	
	8.	Write applications of human genome project in medical sciences.	
	9.	Define: Pharmacogenetics and Pharmacogenomics	
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Q-3	· (a)	Write a note on Sanger's method of sequencing of DNA.	(0.0)
	(b)		(06)
		OR	(06)
	(b)	 Mention various positional dependent strategies. Write short note on chromosome walking. 	(03+03)
Q – 4	(a)	Write a note on Diabetes Mellitus.	(06)
	(b)	Discuss various cancer causing genes.	(06) (06)
		OR	(00)
	(b)	"Cystic fibrosis is a monogenic but multisystem disease" Justify the statement.	(06)
Q-5	(a)	What is Mucopolsaccharidosis? Explain any two types of diseases.	(06)
	(b)	Write short notes on the following: 1) Phenylketonuria 2) LHON OR	(03+03)
	(b)	What is sphingolipodystrophy? Diagramatically explain sphingolipodystrophy associated with metabolism of ganglioside, sphingomyelin and sulfatids.	(06)
Q – 6	(a)	Enlist the methods for detection of known mutation and explain any one in detail.	(06)
	(b)	Write a note on effect of drugs in genetic variation.	(06) (06)
		OR	(00)
	(b)	Explain SSCP and DGGE techniques for the scanning of genes for unknown mutation.	(06)
	-	X	