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## SARDAR PATEL UNIVERSITY

M.Sc. (Integrated Biotechnology) Tenth Semester Examination Monday, 18<sup>th</sup> March, 2019

## PS10CIGMB1 - Human Genetics

10:00 a.m. to 01:00 p.m.

		Total marks: 70		
	Choose the most appropria alternatives given:	te answer from the four (08)		
i	Construction of comprehensive map for the first time in 1987 was			
	based on			
	a) RFLP	c) Minisatellite		
	b) STS	d) VNTR		
ii	In Maxam-Gilbert sequencing formic acid is used			
	a) for depurination	c) for methylation		
	b) for hydrolyses of pyrimidines	d) none of above		
iii	is considere	ed to have an autosomal dominant		
	pattern of inheritance in which a person are born with one mutated			
	copy of the NFI gene in each cell			
	a) Mucoviscidosis	c) Huntington's chorea		
	b) Recklinghausen Disease	d) None of above		
iv	Which one of the following is not a subtype of Diabetes mellitus?			
	a) Type I Diabetes	c) Type I Diabetes		
	b) MODY	d) Gestational diabetes		
v	An organism with partial defici	ency of melanin synthesizing enzymes		
	or reduced level of melanin is known as			
	a) Albino	c) Albinoid		
	b) Achromasia	d) Achromatosis		
vi	is caused by a mu	itation in a gene those results in the		
	accumulation of homogentisic ac	eid (HGA).		
	a) Lipidosis	c) Phenyl ketonuria		
	b) Alkantonuria	d) None of above		
vii	is a mutation of	single nucleotide resulting in a codon		
	that codes for a different amino	acid.		
	a) Nonsense mutation	c) Neutral mutation		
	h) Missense mutation	d) Silent mutation		
vii	i Antimalarial drug induced hen	nolytic anemia in African males due to		
	the deficiency of Glucose 6 phosphate dehydrogenase is an example of			
	variation affecting			
	a) Phase I metabolism	c) Pharmacodynamic response		
	b) Pharmacokinetic response	d) Phase II metabolism		

Q.2	Answer any SEVEN from the following:			
	i.	Write applications of pulse field gel electrophoresis.	(14)	
	ii.	Differentiate between Physical markers and Genetic markers.		
	iii.	Describe the causes of Royal disease.		
	iv.	What is BMI?		
	ν.	Classify disorders of inborn error of metabolism.		
	vi.			
	vii.	Name human Mitochondrial syndromes.		
	viii.	Define: Ecogenetics and Pharmacogenomics.		
	ix.	Write significance of DNA testing.		
Q.3	(a) (b)	Enlist physical makers used for mapping. Explain clone map in detail.  Write a detail note on Chromosome walking.  OR	(06) (06)	
	(b)	Explain any two positional independent strategies used for mapping human disease gene.	(06)	
Q.4	(a)	Write a note on Oncogenes.	(06)	
	(b)	Describe in detail genetic basis of Huntington's disease.  OR	(06)	
	(b)	Write a notes on the following:  (i) Neurofibromatosis  (ii) Genes associated with IDDM and NIDDM	(3+3)	
Q.5	(a)	Describe the causes, symptoms and mechanism of Maple syrup urine disease.	(06)	
	(b)	Describe genetic aspects of PKU and Pome disease.	(06)	
	(b)	OR Give an overview of sphingolipidosis.	(06)	
Q.6	(a)	Enlist the techniques used for detection of known mutation. Explain any 2 of them in detail.	(06)	
	(b)	Describe ethical issues in medical genetics.  OR	(06)	
	(b)	<ul><li>(i) Write a note on Conjugation reaction</li><li>(ii) Enlist techniques for the scaning of genes for unknown mutations.</li></ul>	(03) (03)	
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