Reg. No															

B.Tech DEGREE EXAMINATION, MAY 2024

Fifth Semester

18BTE420T - HUMAN GENETICS

(For the candidates admitted during the academic year 2018 - 2019 to 2021 - 2022)

Note:

i. **Part - A** should be answered in OMR sheet within first 40 minutes and OMR sheet should be handed over to hall invigilator at the end of 40th minute.

ii. **Part - B** and **Part - C** should be answered in answer booklet.

Time: 3 Hours **Max. Marks: 100**

PART - A (20 × 1 = 20 Marks)

Answer **all** Questions

		Marks	BL	CO
1.	Prader Willi syndrome is due to (A) absence of 15th chromosomal pair (B) absence of maternal 15 chromosome (C) absence of paternal 15th chromosome (D) absence of telomere in 15th chromosome	1	1	1
2.	One gene controlling multiple traits independently is known as_____ (A) Genetic heterogeneity (B) Mosaicism (C) Pleiotropy (D) Multiple allele	1	1	2
3.	_____ is defined as the proportion of symptomatic individuals that carry a particular mutation in a population (A) expressivity (B) heterogeneity (C) onset (D) penetrance	1	1	2
4.	How many Barr bodies are present in a female with Turner syndrome (A) 1 (B) 2 (C) 3 (D) 0	1	2	2
5.	If the centromere is near the end, the chromosome is called (A) Telocentric (B) metacentric (C) submetacentric (D) acrocentric	1	1	3
6.	Which one of the following are structural proteins in the nucleus? (A) Heterochromatin Proteins (B) Polycomb group (PcG) proteins (C) Histones (D) DNA polymerases	1	1	3
7.	The repeat sequences in the genome with more copy numbers are known as _____ (A) SINEs (B) LINEs (C) DNA transposons (D) LTR Elements	1	2	3
8.	RNAi denotes which one of the following (A) RNA Induction (B) RNA Insertion (C) RNA Inference (D) RNA Interference	1	3	3
9.	Which of the following is an example of monosomy? (A) 46,XX (B) 47,XXX (C) 22, X (D) 45,X	1	3	4
10.	In a Robertsonian translocation fusion occurs at the (A) centromere (B) telomere (C) Long arms (D) Short arms	1	3	4

11.	The unit of physical mapping is (A) Mu (B) cM (C) bp (D) Mu/cM	1	3	4
12.	Which of the following foetus will survive? (A) monosomy 18 (B) polyploid (C) monosomy 21 (D) monosomy X	1	3	4
13.	What is the reason for Mendel not recognizing linkage phenomenon in his experiments? (A) He studied only pure plants (B) Characters he studied were situated in different chromosomes (C) He did not have a powerful microscope (D) Characters he studied were situated in same chromosomes	1	5	5
14.	Percentage of crossing over is more when _____ (A) genes are located on different chromosomes (B) genes are not linked (C) linked genes are located close to each other (D) linked genes are located far apart from each other	1	5	5
15.	Which of the following is a X linked recessive disease? (A) Duchenne muscular dystrophy (B) Cystic fibrosis (C) Bronchio-oto-renal syndrome (D) Crohn's Disease	1	4	5
16.	Ten mapping units are equal to _____percent of recombination frequency. (A) 1 (B) 10 (C) 100 (D) 0.1	1	5	5
17.	Amniocentesis can be done between _____ weeks with minimal risk. (A) 6 -7 (B) 14 - 20 (C) 20 - 25 (D) 25-30	1	5	6
18.	CVS is collected from the _____ (A) placenta (B) uterus (C) endometrium (D) fetus	1	4	6
19.	Commonly used method to detect the deletion of DNA sequence is_____ (A) ligation-dependent probe amplification (B) Multiplex primer PCR assay (C) ARMS PCR assay (D) oligonucleotide hybridization assay	1	4	6
20.	For a proper pedigree tree, genetic counselors need information of at least _____ generations (A) 2 (B) 5 (C) 3 (D) 4	1	5	6

PART - B (5 × 4 = 20 Marks)

Answer **any 5** Questions

		Marks	BL	CO
21.	Write a brief note on pleiotropy with a suitable example.	4	1	1
22.	Define the term heterogeneity with a suitable example.	4	1	2
23.	Write a brief report on rRNA genes in the human genome.	4	1	3
24.	Write a brief note on mixoploidy.	4	1	4
25.	Discuss the clinical manifestations of CFTR disease.	4	3	5
26.	Give a brief note on various types of genetic markers in the human genome.	4	4	5
27.	What are the determinants of personalized medicine?	4	4	6

		Marks	BL	CO
	PART - C (5 × 12 = 60 Marks) Answer **all** Questions			
28.	(a) Write an essay on monogenic inheritance pattern, characteristic, pedigree, segregation with examples.	12	3	6
	(OR)			
	(b) Explain X-linked inheritance and explain with one genetic disorder of X-linked inheritance.			
29.	(a) Elaborate the organizational of human genomic DNA with suitable diagram.	12	4	5
	(OR)			
	(b) Differentiate coding and non-coding genes with its significance.			
30.	(a) How chromosome instability is analyzed? Write their clinical features.	12	2	4
	(OR)			
	(b) Explain how cohorts and trio studies are associate with GWAS.			
31.	(a) How genetic mapping is done? Explain the procedure.	12	1	3
	(OR)			
	(b) Write a note on positional cloning? Elaborate on techniques used to identify disease genes.			
32.	(a) Write an essay on genetic testing. Elaborate on population carrier analysis with examples.	12	2	2
	(OR)			
	(b) Write an account on UPD with suitable examples.			

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