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B.Tech DEGREE EXAMINATION, NOVEMBER 2023

Fifth Semester

18BTE420T - HUMAN GENETICS

(For the candidates admitted during the academic year 2020 - 2021 & 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.	_____ is a condition where two dominant alleles are expressed equally when present in the heterozygous state. (A) Co-dominance (B) Disomy (C) Phenocopy (D) Variable expressivity	1	1	1
2.	Haemophilia is more common in males because it is a ___condition. (A) Y - linked dominant (B) X linked recessive (C) Mitochondrial inherited (D) X linked dominant	1	1	1
3.	The observed manifestation and effect of the action of a gene or genes is called____. (A) Phenocopy (B) Pleiotrophy (C) Phenotype (D) Genotype	1	2	1
4.	When a human has a mutation in BRCA1 or BRCA2 gene, then (A) 100% the person develops breast cancer, as BRCA1/2 gene is the main cause (B) may or may not be the person develops breast cancer, as there are other external factors (C) the person does not develop breast cancer, as BRCA1/2 is not responsible for breast cancer (D) it will only affect females carrying the mutation, not males	1	1	2
5.	The normal human chromosome diploid number is (A) 23 (B) 24 (C) 46 (D) 48	1	1	2
6.	Normally, endogenous non-coding dsRNAs that bind to mRNAs and inhibit their translation is ---------- (A) RISC (B) ShRNA (C) miRNA (D) mRNA	1	1	2
7.	DNA sequence that can change its position within genome is called---------- (A) Transcription start site (B) Promoter region (C) transposable element (D) Introns	1	1	2
8.	The most abundant macromolecule of chromatin is _____ (A) protein (B) DNA (C) RNA (D) glycoprotein	1	1	3
9.	The number and appearance of chromosomes in an organism is called (A) ideogram (B) karyogram (C) karyotype (D) . idiotype	1	1	4
10.	The chromosome that is not sub-metacentric is (A) X (B) 15 (C) 17 (D) 18	1	1	5

11.	XIST and TSIX are (A) complementary to each other (B) protein coding genes (C) mitochondrial genes (D) Are analogous regions	1	4	4
12.	Which of the following is a fully equipped transposon? (A) LTR element (B) DNA transposon (C) SINES (D) LINES	1	4	4
13.	Percentage of crossing over is more when (A) Genes are located in a 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	1	5
14.	How many linkage groups are there in an organism containing 5 metacentric chromosomes? (A) 0 (B) 6 (C) 10 (D) 5	1	4	5
15.	Which of the following is a non-PCR marker? (A) RAPD (B) RFLP (C) SSR (D) VNTR	1	4	5
16.	1cM corresponds to _____recombinant fraction (A) 0.1 (B) 0.01 (C) 0.001 (D) 1	1	4	5
17.	_____is the study of the role of the genome in drug response. (A) Functional genomics (B) Comparative Genomics (C) Pharmacogenomics (D) Structural genomics	1	1	5
18.	Amniocentesis is a method for (A) Inducing abortion (B) Artificial insemination (C) Determining amino acid sequence (D) finding abnormalities in fetus	1	4	5
19.	Which of the following condition is caused by DNA repair mechanism impairment--- (A) Fragile X syndrome (B) Fanconi's anemia (C) Fredric's syndrome (D) Prader-Willi syndrome	1	1	5
20.	Which type of the blood cell is affected in sickle cell disease? (A) WBC (B) RBC (C) Platelets (D) Plasma	1	1	5

PART - B (5 × 4 = 20 Marks)

Answer **any 5** Questions

		Marks	BL	CO
21.	Cystic fibrosis is a condition with a frequency of 0.01 in a population. Calculate the following: i) the frequency of the recessive allele in the population ii) the frequency of the dominant allele in the population iii) the percentage of heterozygous individuals (carriers) in the population.	4	4	2
22.	Write an account on the genetic causes of Angelman syndrome.	4	1	1
23.	Define satellite DNA. List down few applications of them.	4	1	2
24.	How do female mammalian cells handle two X chromosomes?	4	5	4
25.	Write a short note on positional cloning.	4	1	5
26.	Define genetic counseling and its applications	4	3	5

		Marks	BL	CO
27.	Mention the genetic etiology of Bronchio-oto-renal syndrome.	4	4	5

PART - C (5 × 12 = 60 Marks)

Answer **all** Questions

		Marks	BL	CO
28.	(a) Discuss in detail the factors responsible for deviations of Mendel's laws.	12	1	1
	(OR)			
	(b) Explain the concepts of penetrance and disease anticipation with suitable examples.			
29.	(a) Write an essay on the non-coding DNA elements in human genome.	12	2	2
	(OR)			
	(b) Write an account on mitochondrial genome organization and its limited autonomy.			
30.	(a) Write a detailed account on the mechanism of X-inactivation.	12	2	3
	(OR)			
	(b) Write an essay on autosomal and allosomal chromosomal abnormalities observed in humans.			
31.	(a) Write an essay on the genetics, clinical features and diagnosis of Crohn's disease.	12	1	4
	(OR)			
	(b) Write a detailed account of molecular genetics of Duchenne muscular dystrophy.			
32.	(a) What is genetic testing?. Explain the types of genetic testing with suitable examples.	12	1	5
	(OR)			
	(b) Write a detailed account on the process and importance of genetic counseling.			

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