LOYOLA COLLEGE (AUTONOMOUS), CHENNAI – 600 034

M.Sc. DEGREE EXAMINATION – **BIOTECHNOLOGY**

SECOND SEMESTER – **APRIL 2022**

PBT 2601 – HUMAN GENETICS

Date: 24-06-2022 Dept. No. Max. : 100 Marks

Time: 09:00 AM - 12:00 NOON

PART – A

Answer ALL the Questions

I. Choose the correct answer **(5 x 1 = 5 Marks)**

1. A man marries a woman, and both do not show any apparent traits of inherited disease. Five sons and two daughters are born, and three of their sons suffer from a disease. None of the daughters are affected. The following mode of inheritance for the disease is
   a) sex-linked recessive b) sex-linked dominant c) autosomal dominant d) none of the above
2. The therapeutic gene takes over the function of a _____ gene
   a) correct b) right c) non-functional d) functional
3. The common gene delivery system for In vivo gene therapy is
   a) Micro injection b) Lipofection c) Adeno viral vectors d) Electroporation
4. Between two chromosomes the exchange of gene is called
   a) Interaction b) Mutation c) Recombination d) Point mutation
5. The main aim of human genome project is
   a) to identify and sequence of all the genes present in the human body
   b) to introduce new genes to human beings
   c) to remove disease causing genes from humans
   d) to improve techniques of finger printing.

II. State whether the following are true or false. **(5x1=5 Marks)**

6. A genetic codon is said to be degenerate or redundant.
7. Mutations that occur in somatic cells are inherited.
8. Spinal muscular atrophy is inherited in an autosomal recessive pattern.
9. Pharmacogenomics deals with the influence of genetic variation on drug response in patients.
10. If female traces are to be assigned to female individuals, ChrX markers yield the same results as AS.

III. Complete the following **(5 x 1= 5 Marks)**

11. Variations in DNA sequence between individuals are termed _____.
12. The technique of direct injection of DNA into skeletal of cardiac muscle is called _____.
13. The biochemical and physiological effects of drugs and the mechanisms of their actions is known as_____.
14. _____ is when an organism has a third copy of a chromosome that should be present in two copies.
15. _____ is a type of structural mutation where a part of chromosomes or a set of genes rotates by 180° on its own axis.

IV. Answer the following within 50 words **(5 x 1 = 5 Marks)**

16. How are ring chromosome formed?
17. What mutation causes Becker muscular dystrophy?
18. What are the problems with gene therapy?
19. Define embryoscopy.
20. Comment on the *SRY* gene.

PART – B

Answer the following each within 500 words. **(5 x 8 = 40 Marks)**
Draw diagrams wherever necessary

21. (a). Discuss on the types of gene mutations.

OR

(b) Write a note on repetitive sequences in human genome.

22. (a) Explain the genetic basis of Duchenne muscular dystrophy.

OR

(b) Discuss on Alzheimer's disease and its syndromes.

23. (a) Elaborate on how organ transplants and stem cell therapy help treat diseases

OR

(b) Explain the role of nucleic acids in combating disease.

24. (a) List the advantages and disadvantages of using DNA vaccines.

OR

(b) Briefly outline the key biochemical and molecular tests that are used in genetic testing.

25. (a) Explain the importance of X chromosome short tandem repeats.

OR

(b) Enumerate the advantages and limitations of Y chromosomal markers.

PART – C

Answer any TWO of the following, each within 1500 words. **(2 x 20 = 40 Marks)**
Draw diagrams wherever necessary.

26. Classify and explain the types of structural chromosomal abnormalities.

27. Explain the pattern of inheritance of genetic disorders.

28. Explain in detail prenatal screening for Down syndrome.

29. Write an essay on the identification of male lineages in forensics.

@@@@@@@