



LOYOLA COLLEGE (AUTONOMOUS), CHENNAI – 600 034

M.Sc. DEGREE EXAMINATION – BIOTECHNOLOGY

SECOND SEMESTER – APRIL 2018

17PBT2ES01- HUMAN GENETICS

Date: 25-04-2018
Time: 01:00-04:00

Dept. No.

Max. : 100 Marks

PART – A

Answer ALL the Questions

I. Choose the correct answer

(5 x 1 = 5 Marks)

- Mutations which cause a complete loss of function of the protein are called
a) Nullisomy b) Null mutations c) Point mutations d) Inversion mutation
- _____ mutations are linked to pachyonychia congenita
a) keratins 6a, 6b, 16, 17 b) keratins 6a, 6b, 15, 17 c) keratins
6a, 6b, 6c, 17 d) keratins 6a, 6b, 16, 18a
- Human immunodeficiency virus (HIV) is a common example of
a) Retrovirus b) Herpes Simplex Virus c) Adeno-associated Virus d) Adenovirus
- Which is the common neural tube birth defect?
a) Encephalocele b) Anencephaly c) Multiple gestation d) Spina bifida
- HumARA forensic DNA markers consist of _____ repeat that is located on the coding region.
a) CCG b) CAG c) GAC d) ACG

II. State whether the following are True or False.

(5x1=5 Marks)

- Mutations that cause a novel function are known as neomorphic mutations.
- Maple syrup urine disease is characterized by the growth of noncancerous tumors called neurofibromas.
- Liposomes trigger a range of immune responses.
- Cordocentesis evaluating fetal metabolism and hematologic abnormalities.
- Genetic ancestry identification is important in forensic cases with no known suspects.

III. Complete the following

(5 x 1 = 5 Marks)

- _____ is the mode of inheritance for Huntington Disease.
- _____ gene codes an enzyme called alpha-1 antitrypsin.
- DNA Vaccine induces _____ immune response.
- _____ is used to visualize the embryo for the diagnosis of structural malformations.
- The _____ Y-SNP marker appears at high frequency almost everywhere in Africa.

IV. Answer the following within 50 words

(5 x 1 = 5 Marks)

- What are large genomic rearrangements?
- How are Mendelian disorders classified?
- What is hematopoietic stem cell?
- Give any two principles used for preventative model.
- What are Y-STRs?

PART B

Answer the following each within 500 words.

(5 x 8 = 40 Marks)

Draw diagrams wherever necessary.

21. (a) Describe the mutations that occur due to the change in the number of chromosomes.

OR

(b) Write short notes on splice site mutations.

22. (a) Outline the pattern of inheritance of genetic disorders.

OR

(b) Write a brief note on neurofibromatosis type 1 and pachyonychia congenita.

23. (a) Explain the mechanism of RNA interference in therapeutics.

OR

(b) Discuss about stem cell therapy.

24. (a) What are the types of genetic testing?

OR

(b) Explain the phases of genetic counseling.

25. (a) Write short notes on the origin of mitochondria and mtDNA.

OR

(b) Write about the advantages and disadvantages of using short tandem repeats?

PART – C

Answer any TWO of the following, each within 1500 words.

(2 x 20 = 40 Marks)

Draw diagrams wherever necessary.

25. Discuss in detail the genetics of neurogenetic disorders.

26. Explain the various gene delivering systems in gene therapy.

27. Give a detailed account on the various noninvasive techniques used for prenatal diagnosis.

28. Explain the identification of male paternity and male geographical origin.

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