



**LOYOLA COLLEGE (AUTONOMOUS), CHENNAI – 600 034**

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

SECOND SEMESTER – APRIL 2017

**16PBT2ES01- HUMAN GENETICS**

Date: 28-04-2017  
01:00-04:00

Dept. No.

Max. : 100 Marks

**PART – A**

**Answer ALL the Questions**

***I. Choose the correct answer*** (5x1 = 5 Marks)

- Identify the cause of Turner's syndrome.  
a) Aneuploidy                      b) Point mutation    c) Non sense    d) Missense
- What is the carrier frequency of cystic fibrosis?  
a) 1/25                      b) 1/50                      c) 1/75    d) 1/100
- Choose the enzyme involved in drug metabolism.  
a) Cytochrome P450                      b) Amylase                      c) Lipase                      d) Caspase
- Which disease is caused by CAG repeat expansion in the gene?  
a) Cystic fibrosis                      b) Alzheimer                      c) Diabetes                      d) Huntington's
- Hair samples can be best analyzed using which marker DNA?  
a) mt DNA                      b) X Chr                      c) Y Chr                      d) Autosomal

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

- Isochromosomes are formed by the longitudinal division of chromosome.
- Diabetes is a multifactorial disorder.
- Adeno-associated virus is used for gene therapy.
- Maternal serum can be used for the prenatal screening of Down syndrome.
- The size of human Y chromosome is 45 Mb.

***III. Complete the following*** (5x1=5 Marks)

- \_\_\_\_\_ % of human genome is coding.
- The number of exons present in DMD gene are \_\_\_\_\_
- SCID stands for \_\_\_\_\_
- Chorionic Villus Sampling (CVS) is done during \_\_\_\_\_ weeks of pregnancy.
- Russian Tsar DNA was analyzed by \_\_\_\_\_ DNA marker.

***IV. Answer the following within 50 words*** (5x1 = 5 Marks)

- Define missense mutation.
- Cite a multifactorial disorder and how is it caused?
- What is pharmacogenetics?
- What is the purpose of prenatal diagnosis?
- Mention the use of a haplotype.

**PART B**

*Answer the following each within 500 words.*

**(5x8 = 40 Marks)**

*Draw diagrams and flowcharts wherever necessary*

21. a) Illustrate the formation of Philadelphia chromosome.

OR

b) Discuss Robertsonian translocation with an example.

22. (a) Enumerate the various applications of Genetics in Medical practice.

OR

(b) Give the logic involved in positional cloning strategy.

23. (a) Discuss gene augmentation therapy used for correcting autosomal recessive disorders.

OR

(b) What are the various tests a donor has to undergo for organ transplantation.

24. (a) Mention the indicators of genetic counseling.

OR

(b) Discuss the indications for invasive prenatal diagnosis

25. (a) What are the ethical considerations of chromosome X testing?

OR

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

**PART – C**

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

**(2 x20 = 40 Marks)**

*Draw diagrams and flowcharts wherever necessary.*

26. Describe the genetics of Duchenne and Becker Muscular Dystrophies.

27. Discuss stem cell therapy.

28. Explain various non-invasive techniques involved in prenatal diagnosis.

29. Explain

i) How mitochondrial DNA analysis is done using sample and reference DNA.

ii) Various repetitive elements in human genome.

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