

MEDICAL GENETICS

PAPER-III

MED. GEN./D/18/53/III

Time: 3 hours

Max. Marks:100

Important Instructions:

- *Attempt all questions in order.*
- *Each question carries 10 marks.*
- *Read the question carefully and answer to the point neatly and legibly.*
- *Do not leave any blank pages between two answers.*
- *Indicate the question number correctly for the answer in the margin space.*
- *Answer all the parts of a single question together.*
- *Start the answer to a question on a fresh page or leave adequate space between two answers.*
- *Draw table/diagrams/flowcharts wherever appropriate.*

Write short notes on:

1. What is meant by enzyme replacement therapy (ERT)? Discuss the parameters of success of ERT in Gaucher disease and compare it with other modes of treatment available for this condition. 2+4+4
2. Mention the types of mutations that you know and discuss how they cause disease. 6+4
3. Name the techniques used for prenatal diagnosis. Mention the prerequisites before performing prenatal diagnosis. Give the advantages and disadvantages of Non-invasive prenatal testing (NIPT) versus invasive techniques. 3+4+3
4. What is Warfarin embryopathy? Explain the mechanism, genes involved and give the differential diagnosis. 2+(2+2+4)
5. Discuss the issues related to Genetic testing and Health Insurance. What is genetic discrimination? What are its social implications? 4+4+2
6. What are triplet repeat disorders? Mention various techniques to detect it. Give an example of one such disorder with its inheritance pattern. 5+2+3
7. What are the databases useful for syndrome recognition? Enlist advantages and disadvantages of each. 6+4
8. Name some diseases with multifactorial inheritance with details of genetics in two of them. Describe the role of Genome Wide Association Screening (GWAS) in identifying candidate genes for multifactorial disorders. 7+3
9. What is meant by Exon skipping? List some of the conditions where it has been experimented and discuss its potential in treating genetic disorders. 5+(2+3)
10. How will you approach a heterozygous missense non-synonymous variant of uncertain significance (VUS) found in clinical exome sequencing in a child with infantile epilepsy? Mention some of the in-silico prediction tools that can be used to decide its pathogenicity. 6+4
