

**MEDICAL GENETICS**

**PAPER-I**

Time: 3 hours  
Max. Marks: 100

MED GEN/D/19/53/I

**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. Define "Gene". Draw the basic structure of a gene. Give some examples of promoter site variations/ mutations and their effects. 2+4+4
2. Give the principle of genomic imprinted disorders. List four examples of such disorders. Describe methods used for diagnosing any one such disorder in detail. 2+4+4
3. What molecular techniques will you apply for finding mutations/ variations in the beta-globin gene? What are the different mutations in beta-globin gene and how do you classify them? What are the areas worldwide and ethnic groups in India where beta-thalassemia and sickle cell disease are prevalent; and what do you think can be the underlying causes? 2+4+4
4. Explain genetic heterogeneity taking "deafness" as a prototype disorder. Discuss various molecular tests which you might require to investigate a deaf child with normal parents and without any other affected family members. 5+5
5. Discuss in brief modes of inheritance in mitochondrial disorders. What is threshold effect or expression in mitochondrial disorders? How will you counsel a couple who have a child with Leigh disease? 2+3+5
6. What do you mean by Mosaicism? Describe the phenomena with diagrams. Name two disorders seen only in mosaic state. What are the salient clinical features which suggest diagnosis of a mosaic disorder? 5+2+3
7. Name the mismatch repair genes and the mechanism of cancer due to the defect in them. How will you counsel an asymptomatic individual with biallelic mutations? 6+4
8. What is consanguinity? Draw a three generation pedigree with a consanguineous marriage in the second generation. Explain with examples, about the genetic disorders, you think is more prevalent in a population where there is consanguinity. 2+4+4

P.T.O.

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9. What is meant by cell free DNA? How can it be used for diagnostic purposes in a genetic clinic? Discuss the advantages and disadvantages of using cell free DNA for diagnosis? 3+3+4
10. What are allelic disorders? Mention about allelic disorders in relation to at least four genes. 2+8

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