

MEDICAL GENETICS**PAPER-I**Time: 3 hours
Max. Marks:100

MED.GEN/D/20/53/I

Important Instructions:

- You are provided with 5 answer sheet booklets. Each individual answer sheet booklet consists of 10 pages excluding the covering jackets.
- Answers to all the questions must be attempted within these 5 answer sheet booklets which must be later tagged together at the end of the exam.
- No additional supplementary answer sheet booklet will be provided.
- 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 Hardy Weinberg Equilibrium in relation to population genetics? What is the difference between genotype frequency and allelic frequency? Describe the factors affects the Hardy Weinberg Equilibrium. 2+5+3
2. Illustrate the differences with suitable examples. 5+5
 - a) Mosaicism and chimerism.
 - b) Penetrance and variable expression.
3. What is OMIM? Name and describe few important online databases and mobile applications used in dysmorphology. 2+8
4. What are the health consequences of consanguinity? Define coefficient of relationship and illustrate this with pedigree analysis. What type of disorders are more common in consanguineous marriages? 2+2+6
5. What are Pseudo-TORCH conditions? Why are they called interferonopathies? Write in detail for any one of them. 4+2+4
6. Define "Gene Expression". Describe the various mechanisms of gene expression. What are the methods for gene expression studies? 2+5+3
7. Explain the mechanism of epigenetics and how do they effect phenotype of genetic disorders. Give one example in detail. 2+5+3
8. a) Enumerate the characteristic feature of mitochondrial inheritance. 5+5
b) Discuss briefly the difficulty and limitations during genetic counselling of a mitochondrial disorders.

P.T.O.

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9. What are the different types of polymorphisms observed in human DNA? Give practical examples of use of STRs (Short Tandem Repeats) in diagnostics. Mention how the knowledge of polymorphism is used in SNP Microarray. 4+3+3
10. Give a brief description of replication of human DNA and how DNA is repaired if damaged. 6+4

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