



**MEDICAL GENETICS**  
**PAPER-I**

GENE/D/17/53 /I

TIME : 3 HOURS  
MAX. MARKS : 100

- 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 question to a question on a fresh page or leave adequate space between two answers.
- Draw table/diagrams/flowcharts wherever appropriate.

Write Short notes on:

- a) What are the different conventional cytogenetic techniques? 3+5+2
  - b) Name some molecular cytogenetic techniques with brief description of each
  - c) What are the advantages of MLPA (Multiplex Ligation-dependent Probe Amplification) and its limitations?
- a) What is Universal New-born Screening (NBS)? 1+4+5
  - b) Which disorders do you think should be included in Universal NBS and why?
  - c) What are the basic laboratory techniques used for NBS for metabolic disorders?
- a) What is multifactorial inheritance? 2+4+4
  - b) Name some disorders included in this group with genetics of each.
  - c) Role of GWAS (Genome-wide association studies) in genetic disorders
- a) How do you recognize X-linked recessive disorders from a pedigree? 3+3+4
  - b) Name some X-linked recessive disorders with a brief description of any two of these.
  - c) What are causes of manifestation of such a disorder in females?
- a) Classify types of mutation. 5+5
  - b) Give examples of each with mechanism underlying such mutations.



6. a) What is hemophilia? 3+7  
b) How do you perform mutation analysis for different types of hemophilia?
7. a) What are animal models? 4+6  
b) Name some animal models used for genetic disorders and mention the advantages for using such models.
8. a) What is the principle of Sanger sequencing? 4+3+3  
b) Mention its advantages over ARMS-PCR in diagnosis of Beta-thalassemia  
c) When will you prefer Sanger-Sequencing over NGS (Next-generation sequencing)?
9. a) What are the various databases you can use for syndrome search? 6+4  
b) How will you use OMIM and what disorders do you look for when you use OMIM?
10. a) What are familial cancers? 4+3+3  
b) Name some familial cancers.  
c) What laboratory technology will you use and how for diagnosis in a family with VHL (Von-Hippel-Lindau disease)?

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