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:

1. 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?

2. 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?

3. 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

4. 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?

5. a) Classify types of mutation.   5+5
    b) Give examples of each with mechanism underlying such mutations.

-**1**-

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   6+4
        search?
     b) How will you use OMIM and what disorders do you look for
        when you use OMIM?

10. a) What are familial cancers?
     b) Name some familial cancers.   4+3+3
     c) What laboratory technology will you use and how for diagnosis
        in a family with VHL (Von-Hippel-Lindau disease)?

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