

**MEDICAL GENETICS**  
**PAPER-II**

GENE/D/17/53/II

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:

- What are spino-cerebellar-ataxias (SCA)? 5+3+2
  - How are different varieties inherited?
  - How do you diagnose the common varieties in the laboratory?
- What are the causes of stroke in young? 4+6
  - Discuss approach to diagnosis of a 26 year male with stroke.
- What are autosomal recessive congenital ichthyosis (ARCA)? 6+4
  - How will you counsel the family of a baby born with ARCA?
- How do you clinically diagnose achondroplasia? 4+2+4
  - How will you confirm the diagnosis by molecular testing?
  - What surveillance protocol and management issues you will discuss with the family?
- What are the common causes of developmental delay in a child? 5+5
  - What will be your counseling issues for prenatal testing in next pregnancy for the family with a child with Down syndrome
- What are the different methods of aneuploidy screening during pregnancy? 6+4
  - Mention the advantages and disadvantages of NIPT (Non-invasive prenatal testing).

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| 7.  | a. What are the causes of recurrent spontaneous abortions in a couple?                                                                               | 6+4 |
|     | b. How will you counsel a couple with one partner being carrier of a 'balanced translocation'?                                                       |     |
| 8.  | a. What are the different soft-markers of aneuploidies detected by fetal ultrasound?                                                                 | 4+6 |
|     | b. How will you investigate for non-immune fetal hydrops?                                                                                            |     |
| 9.  | a. What are the different invasive methods for prenatal testing?                                                                                     | 3+7 |
|     | b. Mention the procedure and indications of each of them.                                                                                            |     |
| 10. | a. What are the different causes of indirect hyperbilirubinemia in a new born?                                                                       | 4+6 |
|     | b. How will you investigate and counsel a couple who has history of a previous baby who died at 2 month of age with jaundice and neurological signs? |     |

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