



**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:

- |    |   |       |
|----|---|-------|
| 1. | a. What are spino-cerebellar-ataxias (SCA)?<br>b. How are different varieties inherited?<br>c. How do you diagnose the common varieties in the laboratory?  | 5+3+2 |
| 2. | a. What are the causes of stroke in young?<br>b. Discuss approach to diagnosis of a 26 year male with stroke.   | 4+6   |
| 3. | a. What are autosomal recessive congenital ichthyosis (ARCA)?<br>b. How will you counsel the family of a baby born with ARCA?   | 6+4   |
| 4. | a. How do you clinically diagnose achondroplasia?<br>b. How will you confirm the diagnosis by molecular testing?<br>c. What surveillance protocol and management issues you will discuss with the family? | 4+2+4 |
| 5. | a. What are the common causes of developmental delay in a child?<br>b. What will be your counseling issues for prenatal testing in next pregnancy for the family with a child with Down syndrome          | 5+5   |
| 6. | a. What are the different methods of aneuploidy screening during pregnancy?<br>b. Mention the advantages and disadvantages of NIPT (Non-invasive prenatal testing).                                       | 6+4   |



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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