

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)?
 - How are different varieties inherited?
 - How do you diagnose the common varieties in the laboratory?5+3+2
- What are the causes of stroke in young?
 - Discuss approach to diagnosis of a 26 year male with stroke.4+6
- What are autosomal recessive congenital ichthyosis (ARCA)?
 - How will you counsel the family of a baby born with ARCA?6+4
- How do you clinically diagnose achondroplasia?
 - How will you confirm the diagnosis by molecular testing?
 - What surveillance protocol and management issues you will discuss with the family?4+2+4
- What are the common causes of developmental delay in a child?
 - What will be your counseling issues for prenatal testing in next pregnancy for the family with a child with Down syndrome5+5
- What are the different methods of aneuploidy screening during pregnancy?
 - Mention the advantages and disadvantages of NIPT (Non-invasive prenatal testing).6+4

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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? | |
