

MEDICAL GENETICS

PAPER-III

Time: 3 hours
Max. Marks:100

MED.GEN/D/20/53/III

Important Instructions:

- *You are provided with 5 answer sheet booklets. Each individual answer sheet booklet consists of 10 pages excluding the covering jackets.*
- *Answers to all the questions must be attempted within these 5 answer sheet booklets which must be later tagged together at the end of the exam.*
- *No additional supplementary answer sheet booklet will be provided.*
- *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 answer to a question on a fresh page or leave adequate space between two answers.*
- *Draw table/diagrams/flowcharts wherever appropriate.*

Write short notes on:

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| 1. Name the biomarkers and their clinical importance in patients with Fabry disease and Gaucher's disease. | 3+7 |
| 2. Antenatal markers for next generation sequencing of Noonan syndrome genes in a fetal sample. | 10 |
| 3. Genome editing in beta hemoglobinopathies. | 10 |
| 4. Whole exome sequencing in newborn screening. | 10 |
| 5. Noninvasive prenatal screening. | 10 |
| 6. Inherited retinal dystrophies and novel therapeutics. | 10 |
| 7. Recent advances in the management of spinal muscular atrophy. | 10 |
| 8. Recent advances in management of mitochondrial disorders. | 10 |
| 9. Enumerate the genetic disorders that can be treated by stem cell transplantation. Discuss anyone one disorder. | 5+5 |
| 10. Preconception care. | 10 |
