 NATIONAL BOARD OF EXAMINATIONS

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:

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

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