MEDICAL GENETICS

PAPER-II

Time: 3 hours MED.GEN/D/20/53/II
Max. Marks:100

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. Approach to diagnosis and investigations of a child with intellectual disability and ichthyosis. 5+5

2. A 4-year-old child with prelingual sensorineural hearing loss is referred to you for genetic evaluation after acquired causes have been excluded. Describe your approach for this child. 10

3. Discuss the management and follow up of a 5-year boy with abnormal behavior of recent onset on a setting of mild to moderate intellectual disability and high homocysteine and urine GC-MS showing a methylmalonate peak. 7+3

4. A newborn screen flags off a high immunoreactive trypsinogen (IRT). The 10 days old boy is referred to you for further evaluation. How will you proceed? 10

5. You have received a report of NGS for an asymptomatic child who has an MRI consistent with a diagnosis of X linked adrenoleukodystrophy. The boy is asymptomatic. There is history of a similar disorder in his maternal aunt's son. He has one maternal aunt who has this one affected boy and one unaffected maternal uncle who does not have children. No other member in the family is affected. The variant is a "variant of unknown significance". Please draw the pedigree with the above available information. How will you further examine to determine the significance of this variant? 10

6. Neuronopathic Gaucher disease – types, clinical and diagnostic considerations. 2+5+3

7. Etiology and counseling for a lady at 18 weeks gestation with bilateral enlarged echogenic kidneys. 5+5

8. A child born to non-consanguineous parents has hemihyperplasia noted at 5 months of age. What is your approach to diagnosis and management? 5+5

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9. How will you approach to counsel a lady with congenital heart disease and history of exposure to warfarin in the first trimester of pregnancy? 10

10. A child with history of photosensitivity over the sun exposed areas with blisters, discoloured teeth and nails is referred to you. Outline your differential diagnosis and approach to diagnosis. 5+5

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