

MEDICAL GENETICS

PAPER-II

Time: 3 hours
Max. Marks: 100

MED GEN/D/19/53/II

Important Instructions:

- 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. What are tumour suppressor genes? List the genes. Describe the mechanism and phenotype caused by any one of the genes listed in detail. 2+2+6
2. A 3-day-old male, who was apparently well in first 2 days after birth, suddenly became unresponsive. What are the basic investigations you will order? Write about approach to a neonate with suspected urea cycle disorder. 3+7
3. What are primary immunodeficiency disorders? How will you approach to investigate a 2-year-old female child with recurrent infections and failure to thrive? 2+8
4. Classify and innumerate few causes of intellectual disability along with dysmorphic features in a child? If you have come across a child with the above features and you are unable to make a provisional diagnosis, how would you explore internet databases to try find out the diagnosis? 5+5
5. What are the causes of male infertility? How would you clinically examine and investigate a couple with infertility? 3+7
6. What do you think can a good test to look for chromosomal anomalies in a "product of conception" (POC) which you have obtained in your laboratory from outside? What are the different chromosomal aberrations you are likely to find in a POC? Mention the various chromosomal aberrations you get in Turner's syndrome. 3+3+4
7. Enumerate the various causes of 46, XX DSD (Disorders of sex development). How will you manage a newborn with congenital adrenal hyperplasia (CAH)? Write about the approach to pre-natal testing for common variety of CAH. 3+4+3
8. How is teratogenicity of a drug determined? Name some infectious agents which can act as teratogens and enumerate their effects. 4+6

P.T.O.

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9. Name some micro-deletion syndromes. How will you investigate and manage a child with Williams syndrome? 5+5
10. What is meant by genetic susceptibility? What are the methods used to study this? Illustrate with examples. 4+6
