

MEDICAL GENETICS

PAPER-I

MED. GEN./D/18/53/I

Time: 3 hours

Max. Marks:100

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. Which are the major mechanisms leading to structural defects in the fetus? What is the difference between a syndrome and an association in dysmorphological diagnosis and give examples of these? 7+3
2. How can a balanced reciprocal translocation carrier have unbalanced gametes? Demonstrate with diagrams. How will you counsel such a person planning pregnancy? 5+5
3. What are the differences between mitochondrial and nuclear DNA? Draw a pedigree showing mitochondrial inheritance. What are the difficulties in counselling a woman with mitochondrial mutations planning a pregnancy? 4+2+4
4. Describe the principle of Sanger Sequencing. In what way it is different from Next generation sequencing? State advantages and disadvantages of each. 4+2+4
5. Explain Knudson hypothesis with examples. What is the relevance of clinical history taking in suspecting familial cancer syndrome in a lady with breast cancer? What are the genes involved in familial breast cancer? 5+3+2
6. What is meant by Hardy Weinberg Equilibrium? Mention situations when this equilibrium is disturbed. 5+5
7. Describe main features of Beckwith-Wiedeman syndrome and mention the mechanisms leading to this disorder. 5+5
8. Elaborate the principle of Multiple Ligation Dependent Probe Amplification (MLPA). In which conditions is it most useful? Give a list in order of priority/situations where it is used in clinical practice. 6+2+2
9. Name X-linked recessive disorders commonly seen in a Genetic Clinic. Describe any one in detail. Describe the mechanisms which leads to a manifesting carrier. 3+5+2
10. Justify the inclusion of various disorders of inborn errors of metabolism in newborn screening. What are the basic laboratory techniques used to detect the newborn metabolic disorders? 5+5
