

**MEDICAL GENETICS**

PAPER-II

MED. GEN./D/18/53/II

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. How will you approach a case of recurrent Non-Immune Hydrops? What are the relevant investigations you will do before planning next pregnancy in this case? 5+5
2. A pregnancy was terminated due to the fetus having lethal skeletal dysplasia. Enumerate the causes of lethal skeletal dysplasias. What is the role of the medical geneticist when such a termination is planned? 6+4
3. Define craniosynostosis. Name the common syndromes with craniosynostosis. Give the relevance of Fibroblast growth factor receptor genes in this disorder. 2+4+4
4. Discuss approach to a baby born with Neural tube defect. Discuss differential diagnosis and management before next pregnancy in a mother with a previous child with neural tube defect. 4+6
5. Give the differential diagnosis of Cherry red spot found in a 2-year-old child and discuss the management of this case. 5+5
6. Describe the molecular basis and pathophysiology of Sickle cell disease. Discuss the clinical features in brief and how will you manage a case with sickle cell crisis. 4+4+2
7. What is meant by first trimester aneuploidy screening? How will you proceed when a lady is found to be positive for Trisomy18 in this screening? 5+5
8. Explain various DNA repair mechanisms that function in a normal cell. How will you counsel a family with the proband diagnosed with Lynch syndrome? 6+4
9. Discuss the possible genetic causes in a child with developmental delay and macrocephaly. Enlist investigations that are to be done before ordering molecular testing. 6+4
10. A newborn is admitted with seizures and hyperammonemia. Give the differential diagnosis of this case. What investigations will you plan in order to reach to the diagnosis. How will you counsel the parents of this neonate? 4+4+2

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