

MEDICAL GENETICS

PAPER-III

Time: 3 hours
Max. Marks:100

MED GEN/D/19/53/III

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. Write about the genetics of spinal muscular atrophy (SMA). What are the various approved and experimental medications to treat SMA and what are their mechanisms of action? Mention about the route and dosage of one such medication. 4+3+3
2. What do you think is the importance of the following statement: "How does cells sense and adapt to oxygen availability"? Tell us about the molecular pathways related to the above and the rationality of the knowledge to treat cancers. 2+4+4
3. What is the Act to prevent female foeticide in India? Do you think such an Act is necessary; what steps you would like to take to prevent female foeticide if you become a policy maker for this? 3+7
4. How will you do prenatal screening of Neural tube defects? How will you approach a couple who had two consecutive pregnancies affected with Anencephaly? 4+6
5. Describe the uses of Dried blood spot (DBS). Briefly describe the technique and precautions. What are the advantages and disadvantages of expanded screening using DBS? 2+2+6
6. What is hematopoietic stem cell transplantation (HSCT)? Mention few examples of HSCT done for non-hematological disorders and their outcome. 4+6
7. What are the various disorders where fetal surgical procedures are tried? Enumerate the principle, success and caveat of one such procedure. 5+5
8. Describe the phenomena of expansion of repeats in causing genetic disorders. What are the modes of inheritance, state with examples and diagram showing the recurrence risk in each type? 3+7

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9. Describe the pathogenesis and types of Gaucher's disease. What are the management options available in current times? 5+5
10. What is plagiarism and how should it be avoided during paper writing? What are the essential steps to be followed while writing a research paper? Write about statistical tools which you would like to use while trying to find the association of certain genetic variation in a population with a disease. 3+3+4
