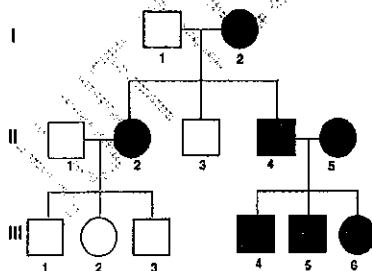


**Note:** (a) All questions are compulsory.

(b) Marks are indicated against each question in square brackets.

(c) The candidate is allowed to make suitable numeric assumptions wherever required for solving problems.

- 1) Describe the differences in the genetic mutations that cause Duchenne and Becker Muscular Dystrophy. How do these differences influence the severity and progression of each condition? Discuss the importance of genetic counseling for families affected by these conditions, especially considering the role of female carriers in the transmission of the disease." [4 Marks] *CO4*
- 2) Given a scenario where a patient presents with early onset of muscle weakness, particularly in the upper arms and lower legs, as well as contractures in the elbows, ankles, and neck with cardiac symptoms and a family history of similar symptoms:
  - a) Diagnose the genetic condition related to muscular dystrophy.
  - b) Discuss inheritance pattern and different mutated genes involved in developing the condition. [4 Marks] *CO4*
- 3) Explain various genetic mutations that have been linked to an increased risk of developing Parkinson's disease. How does the mutation potentially contribute to the development of the disease? [4 Marks] *CO1*
- 4) What is Quad screening? How does it help the physicians and Genetic Counselors? [4 Marks] *CO1*
- 5) Discuss Invasive and Non Invasive Prenatal Diagnostic Tests and indications when these will be used? [3 Marks] *CO2*
- 6) Being Intersex is not a disorder, disease or condition, Comment upon the statement in light of available literature and research findings. [3 Marks] *CO2*
- 7)



The pedigree shows the presence of attached earlobes (autosomal recessive trait) through a family's generations. Having attached earlobes is an autosomal recessive trait. If individuals I-1 and I-2 had a fourth child, what is the chance that the child would have attached earlobes? Though recessive, why does this recessive trait is appearing in each generation? [3 Marks] *CO4*