

**MEDICAL GENETICS**  
**PAPER-III**

GENE/D/17/ 53 /III

TIME : 3 HOURS  
MAX. MARKS : 100

- 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 question to a question on a fresh page or leave adequate space between two answers.
- Draw table/diagrams/flowcharts wherever appropriate.

Write Short notes on:

- What are lysosomal storage disorders (LSD's)?
  - What is ERT (Enzyme-replacement therapy) for LSDs?
  - What are novel strategies/ other strategies to treat LSDs?4+3+3
- What are the pros and cons of stem cell banking?
  - How will you counsel a couple whose previous baby has beta-thalassemia and now they have come to get your advice for stem cell banking in future pregnancies?5+5
- What are SiRNAs and their mechanisms of action?
  - What is the role of SiRNA in therapeutics?5+5
- What are the different principles and methods used for NGS (Next- generation sequencing)?
  - Make a flow chart showing principle of NGS data analysis.
  - When will you prefer Exome sequencing by NGS over Sanger Sequencing for single gene disorders?3+3+4
- What are the different 'in-silico' tools used for pathogenicity testing of a sequence variation?
  - What are the guidelines you will follow and how will you denote the pathogenicity of sequence variation?6+4
- What are different gene-transfer vehicles used in gene therapy?
  - Name some disorders amicable to gene therapy.6+4

7. a. How will you investigate a child with suspected DMD (Duchenne muscular dystrophy) 6+4  
b. What are the recent treatment strategies for DMD?
8. a. What is pharmacogenetics? 2+2+2+4  
b. How do genes affect the metabolism of Warfarin?  
c. What are the fetal effects of maternal warfarin intake?  
d. Name some teratogenic drugs and mention their effects on fetus.
9. What are the functional studies used to detect pathogenicity of a novel sequence variation detected in a single gene disorder? 10
10. a. What is osteogenesis imperfecta (OI)? What are the inheritance patterns? How will you investigate? 6+4  
b. How will you treat a child with OI?

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