

**M. Sc. (Medical Biotechnology) Sem-II (Choice Based Credit System) :**  
**WINTER - 2018**  
**SUBJECT: MEDICAL GENETICS**

Day : Wednesday  
Date : 24/10/2018

**W-2018-1295**

Time : 10.00 AM TO 01.00 PM  
Max. Marks : 60

**N. B. :**

- 1) **Q. No. 1 and Q. No.5 are COMPULSORY.** Out of remaining questions attempt **ANY TWO** questions from each section.
- 2) Answers to both the sections should be written in the **SEPARATE** answer books.
- 3) Draw neat and labeled diagram **WHEREVER** necessary.
- 4) Figures to the right indicate **FULL** marks.

**SECTION - I**

- Q.1** Attempt the following in brief (**Any FIVE**) (10)
- a) Define autosomal recessive giving one example.
  - b) State the genetic abnormality in sickle cell anemia.
  - c) Define monosomy giving one example.
  - d) What are oncogenes?
  - e) Define frame shift mutation.
  - f) Define epistasis giving example.
- Q.2** Attempt the following : (10)
- a) Explain law of segregation with an example.
  - b) Define pleiotropy. Illustrate with an example.
- Q.3** Attempt the following : (10)
- a) Write a note on Rh factor with regards to its role in blood transfusion.
  - b) Explain the structure of human chromosome with well labeled diagram and state its classifications on the basis of the position of centromere.
- Q.4** Write short notes on **ANY TWO** of the following : (10)
- a) X-chromosome inactivation
  - b) Birth defects
  - c) Haemophilia
  - d) Mitochondrial inheritance

**SECTION - II**

- Q.5** Attempt **ANY TWO** of the following : (10)
- a) Describe the genetic abnormality and symptoms of Down's syndrome.
  - b) What is karyotype? Describe the procedure used for its analysis.
  - c) Describe various causes for development of cancer.
- Q.6** Attempt the following : (10)
- a) What is the cause for XXY syndrome? What are the symptoms?
  - b) What is SRY? Explain its role in sex determination.
- Q.7** Attempt the following : (10)
- a) Describe the principle of fluorescent *in situ* hybridization technique and state its applications.
  - b) What is pre-natal diagnosis? Explain its significance.
- Q.8** Give an account on various chromosomal abnormalities giving suitable example. (10)

**OR**

Describe gene therapy giving suitable examples.