M. SC. (MEDICAL BIOTECHNOLOGY) SEM-II (CHOICE BASED CREDIT SYSTEM): WINTER - 2017

SUBJECT: MEDICAL GENETICS

10.00 AM TO 01.00 PM Day Time: : Tuesday Date : 31/10/2017 Max. Marks: 60 W-2017-1051 N.B.: 1) Q.No.1 and Q.No.5 are COMPULSORY. Out of remaining questions attempt **ANY TWO** questions from each section. Answers to both the sections should be written in **SEPARATE** answer books. 2) Figures to the right indicate **FULL** marks. 3) SECTION - I **Q.1** Define **ANY FIVE** of the following terms: [10] a) Loss of function mutations. b) Autosomal recessive. c) Dosage compensation. d) Chromosomal mosaicism. e) Triploidy. f) Monosomy. g) Test cross. Q.2 Attempt the following: [10] a) What is codominance? Explain with an example. b) What are multiple alleles? How they affect the expression of blood groups? Attempt the following: Q.3 [10] a) Explain genetic linkage giving suitable example. **b)** What is extrachromosomal inheritance? Write short notes on **ANY TWO** of the following: **Q.4** [10] a) Jacob's syndrome b) Law of independent assortment c) Cystic fibrosis SECTION - II Q.5 Attempt **ANY TWO** of the following: [10] Write the cause and symptoms of Turner's syndrome. **b)** Describe the inborn errors of lipid metabolism. c) Explain the role of tumor suppressor proteins in control of cell cycle. Attempt the following: [10] **Q.6** a) Give the etiology and symptoms of disease hemophilia. b) Outline the extensions of Mendelian genetic principles. **O.**7 Attempt the following: [10]a) What is amniocentesis? For what purpose it is used? b) What is X – Chromosome inactivation? Explain its significance. [10] Write **ANY ONE** of the following: **Q.8** a) Describe the structure of human chromosome. Classify human chromosomes on the basis of centromere position. b) Describe various techniques used for diagnosis of chromosomal abnormalities.

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