

COURSE CODE: 10B1WBT838

MAX. MARKS: 15

COURSE NAME: Genetic Counseling

COURSE CREDITS: 03

MAX. TIME: 1 HRS.

*Note: All questions are compulsory. Carrying of mobile phone during examinations will be treated as case of unfair means.*

Q1(a) A person of 20 years age visited you as genetic counselor; his father was suffering from Huntington diseases and mother was normal. a) Prepare a query sheet to identify his chances of getting the disease. b) What is the basis of this disease and suggest predictive genetic testing for him. c) What are the implications linked to a positive testing for him.

(b) How does a genetic counselor identify the X-linked pedigree pattern? [3+2 Marks]

Q2(a) Albinism in humans is inherited as an autosomal recessive trait. For the following families determine the genotypes of the parents and offsprings. When two alternate genotypes are possible, list both and provide reasons. a) Two normal parents have five children, four normal and two albinos. b) A normal male and albino female have six children, all normal.

(b) If two parents, both heterozygous and suffering from an autosomal dominant disorder, have four children, what is the probability that two are normal?

(c) You are a genetic counselor and a couple visits your clinic to know – what is the probability that in a family of three children, all are females? [2+2+1 Marks]

Q3(a) A boy with Klinefelter syndrome (47, XXY) is born to a mother who is phenotypically normal and a father who has X-linked skin condition. The mother's skin is completely normal with no signs of abnormality. In contrast her son has patches of normal skin and patches of abnormal skin. a) Which parent contributed the abnormal gamete? b) Using the appropriate genetic terminology, describe the meiotic mistake that occurred. Be sure to indicate in which division the mistake occurred?

(b) What is the reason of Down's syndrome? Elaborate the symptoms associated with this disorder. How does genetic counseling help the patient and the family members?

(c) "Human genetic research and genetic counseling were discredited by Eugenics." Justify the statement. [2+2+1 Marks]