

JAYPEE UNIVERSITY OF INFORMATION TECHNOLOGY, WAKNAGHAT

TEST -2 EXAMINATION- APRIL-2023

COURSE CODE (CREDITS): 18BJWBT 831, 03

MAX. MARKS: 25

COURSE NAME: Genetic Counselling

COURSE INSTRUCTORS: Prof. Sudhir Kumar

MAX. TIME: 1 Hour 30 Minutes

*Note: All questions are compulsory. Marks are indicated against each question in square brackets.*

Q1: a) What will be the variations in planning a genetic counselling session for a (male) suffering from – a) only autosomal recessive disorder b) only autosomal dominant disorder c) only X-linked dominant disorder d) only X-linked recessive disorder e) only Y-linked disorder. Prepare a query sheet/questionnaire. CO2 [04]

b) Can we eliminate Bardet Biedl Syndrome from human population? Explain your answer. CO2 [01]

Q2: a) Mention the major types of Muscular Dystrophies – their cause, symptoms, diagnosis and treatment options. What support and help is extending to patients by Muscular Dystrophy Association of Solan, Himachal Pradesh. CO3 [03]

b) “Life in a bubble – SCID implications!” Comment upon the statement in light of testing, treatment and counselling. CO3 [02]

Q3: a) List out the origin and complications linked to the individuals with the following conditions – XXY, XO, XYY. CO4 [03]

b) A woman is having her niece (sister’s daughter) with trisomy of 21 chromosome. Predict her chances to give birth to a child with same condition. Justify your answer. CO4 [02]

Q4: a) A population that is in Hardy-Weinberg equilibrium, 26% of the individuals are recessive homozygous for a certain condition. In a population of 12,500, calculate the percentage of dominant individuals and heterozygous individuals. CO1 [03]

b) “Affected man will have affected sons” Is it a true statement? Support your answer with scientific reasoning CO1 [02]

Q5: a) Mention the prenatal tests available now and pros and cons linked with each. CO4 [03]

b) Create a hypothetical pedigree of at least 03 generations of a girl suffering from colour blindness. Girl – as proband may be assumed in any of the generations. CO4 [2]