

JAYPEE UNIVERSITY OF INFORMATION TECHNOLOGY**T2 Examination - April 2018****B.Tech (8th Semester)****Course Code: 11B1WBT838****Max. Marks: 25****Course Name: Genetic Counseling****Max. Time: 1Hr. 30Min.****Course Credit: 03**

All questions are compulsory. Be precise in answers.

1. Explain followings:- (5 X 2 =10)
 - a. Discuss read-through mutations and non-sense mutations. Discuss their examples also.
 - b. Give detailed procedure of karyotyping along with its advantages.
 - c. Name the disease in which Guthrie test is performed. How is this test performed?
 - d. Directive and non-directive genetic counseling.
 - e. What are advantages and disadvantages of pre-symptomatic genetic testing? Discuss with an example.
2. Based on symptoms, 'loss of perception of reality, hallucination and delusion' identify this disease. What are various factors that cause this disease? (3)
3. What are differences between Duchenne muscular dystrophy and Becker muscular dystrophy?(3)
4. Identify a disease caused due to CAG repeat expansion. What are its symptoms and discuss role played by CAG repeats in the susceptibility of this disease? (3)
5. What are various genes associated with Alzheimer's disease. Give their chromosomal location and associated phenotype. (3)
6. Discuss briefly BAP and TAU hypotheses for Alzheimer's disease. (3)