

JAYPEE UNIVERSITY OF INFORMATION TECHNOLOGY

T3 EXAMINATION, 2015

B.TECH – Bioinformatics (Semester VIII)

COURSE NAME: Genetic Counseling

MAX.MARKS:35

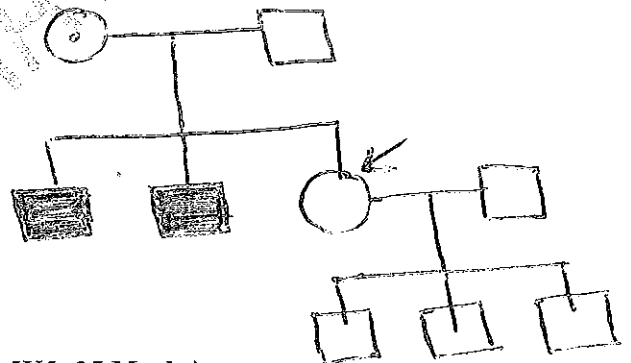
COURSE CODE: 11B1WBT838

MAX.TIME: 2 HRS

 Note: Carrying of mobile phones during examinations will be treated as a case of unfair means.

Section A (2 Marks each; 2X5=10 Marks)

1. What are β -amyloid plaques and neurofibrillary tangles and how are these formed in Alzheimer's disease.
2. Name various genes involved in predisposition to Alzheimer's disease, their chromosomal location and phenotype they show.
3. What do you understand by penetrance and empirical recurrence risk?
4. What are clinical features of Bloom's syndrome?
5. Calculate posterior probabilities of a proband indicated in the following figure of being carrier or non-carrier.



Section B (5 marks each; 5X5=25 Marks)

6. What is targeted killing of specific cells in gene therapy? Explain with an example.
7. Explain BAP and TAU hypotheses for Alzheimer's disease.
8. Write a note on (a) *in-vivo* and *ex-vivo* gene therapy, and (b) HUGO ethics committee statement on gene therapy research.
9. Discuss hereditary colorectal cancer syndromes (a) hereditary non-polyposis colorectal cancer, and (b) familial adenomatous polyposis wrt their epidemiology, symptoms and screening methods for their detection.
10. Explain various beneficiaries and elements of genetic counselling.