

JAYPEE UNIVERSITY OF INFORMATION TECHNOLOGY, WAKNAGHAT
MID SEMESTER EXAMINATION – 2015
B.Tech Semester – VIII

COURSE CODE: 11B1WBT838
COURSE NAME: GENETIC COUNSELING

MAX. MARKS: 30
MAX. TIME: 2 HRS

Note: All questions are compulsory.

Marks

Section A

(6 X 1 = 6)

1. Give treatment options for Turner's syndrome?
2. What is quad screen test?
3. Differentiate inversion and translocation.
4. How is amniocentesis performed?
5. Name high risk groups that should undergo triple screen test.
6. Differentiate mis-sense and non-sense mutations and give examples.

Section B

1. What is directive and non-directive genetic counseling? (2)
2. Identify an autosomal recessive disease that leads to formation of thick mucus and around 70% of the cases of this disease are due to single amino acid deletion. Give five different functional classes of this disease. (2)
3. What are your suggestions, as a genetic counselor, to the parents who have a Down's syndrome child? Should they plan for the second child? (2)
4. Name genes/chromosomes affected in following disorders:- (3)
 - a. Cystic fibrosis
 - b. Phenylketonuria
 - c. Klinefelter's syndrome
 - d. Patau's syndrome
 - e. Edward's syndrome
 - f. Down's syndrome

Section C

1. How phenylketonuria is diagnosed and what are its symptoms? (3)
2. Discuss epidemiology, symptoms, diagnosis and treatment options for Bardet-Biedl syndrome. (4)
3. Explain 5 major classes of Familial hypercholesterolemia caused due to mutations in LDL receptor. (3)
4. What are various diagnosis and management options for G6PD? (3)
5. What are various problems associated with Patau's syndrome? (2)