**PARVATIBAI CHOWGULE COLLEGE OF ARTS & SCIENCE, MARGAO-GOA**

**3rd Continuous Assessment (CA) (Semester I) – 10th October, 2017**

**Class: S.Y.B.Sc. Biotechnology Paper: BIO-III.E-2 Molecular Genetics**

**Duration: 60 minutes Max marks: 15**

***Instructions:***

***Figures to the right indicate maximum marks to the question.***

1. You work in a genetic counsellor’s office and a man, Paul (58y) has come to you to seek your advice about the possibility of a hereditary disease in his family. While taking a family history, he informs you that his father developed symptoms of a neurological disorder at the age of 55y and died at the age of 68y. Paul’s aunt (his father’s younger sister) recently died at the age of 80y but of natural causes. He also remembers that his grandmother (his father’s mother) had had a similar disease to his father (onset at around the age of 57y) wherein her movement was affected and she also suffered from anxiety, depression and memory loss. Paul is married to Linda and they have two children – Simon and Ellie who are 25y and 23y respectively. Paul is concerned about this disease and wants to know whether he has the risk of developing the disease and whether that risk will be passed on to his children.

The general age of onset of the disease is 50 years and it shows a penetrance of 80%.

1. Tarun and Anita (both aged 35y) have come to seek genetic counselling. They already have two children – a boy (6y) and a girl (4y) but are now concerned because Tarun’s mother recently told them about a daughter she had had before Tarun and his older brother who died of a haemoglobin-related disease with symptoms of jaundice, anaemia, enlarged spleen and liver and misshapen bones. Anita has a younger brother but there does not seem to be a history of the disease in her family. Anita is already pregnant with their third child and they want to know the risk of this child being affected by this disease.

The population carrier risk is 1 in 70.

1. A couple, John and Millie (aged 33y and 32y respectively) have approached you as a genetic counsellor as they are planning to start a family but are concerned about a genetic disease in Millie’s family. Her mother’s younger brother was affected by a muscle wasting disease, with weakness in his arms and legs and died at the age of 18y. Millie has an older sister, Mary (34y) who is married to Andrew and a younger brother, Alan (30y) who is married to Tara. Andrew and Mary have one daughter. After establishing the family history, you advise Millie to have a serum creatine kinase (SCK) test done which she agrees to and her results are negative. The couple are keen to find out the risk of having a boy affected by the same muscle wasting disease as Millie’s uncle.