(b) Who elucidated the structure of DNA? Compare and contrast the features of A, B and Z DNA. (9,9)

5. (a) What is genomic imprinting? Explain the role of genomic imprinting in human disorders taking suitable examples. (9)

(b) State True or False. Justify your answer (any 3) (3×3=9)

(i) Osteoarthritis is an autoimmune and inflammatory disease.

(ii) Cancer is a multifactorial disease.

(iii) Sickle cell anemia is prevalent in regions where malaria is common.

(iv) Inversions can not lead to an abnormality as there is no net loss or gain of genetic material.

6. (a) Discuss the structural features of a chromosome. How are chromosomes classified on the basis of the location of centromere? Explain with illustrations.

(b) What is prenatal testing? Describe in detail any one method each of invasive and non-invasive prenatal testing. (9,9)

[This question paper contains 4 printed pages.]

Your Roll No.....

Sr. No. of Question Paper : 3297 **I**

Unique Paper Code : 3184002005

Name of the Paper : Understanding Genetic Basis of Diseases (GE)

Name of the Course : **B.Sc. (Hons) Biomedical Science (NEP)**

Semester : V

Duration : 3 Hours Maximum Marks : 90

Instructions for Candidates

1. Write your Roll. No. on the top immediately on receipt of this question paper.
2. Attempt **five** questions in all.
3. **Question No. 1** is compulsory.
4. Attempt subparts of a question together.
5. Give illustrations and examples wherever required.

1. (a) Briefly explain the following (any 3) (3×2=6)

   (i) Phenotype

   (ii) Frameshift mutations

   (iii) Consanguinity

   (iv) Euchromatin

   (v) Anaphase lagging

   (b) Differentiate between the following (any 3) (3x3=9)

   (i) Transversion and Transition

   (ii) Mutation and Polymorphism

   (iii) α and β Thalassemia

   (iv) Isodisomy and Heterodisomy

   (c) Fill in the blanks (any 3) (3x1=3)

   (i) A disorder due to deletion of a segment on the short arm of chromosome 5 is _____ .

   (ii) A person with Patau syndrome has a trisomy of chromosome _____ .

   (iii) An example of a sex linked recessive disorder is _____ .

   (iv) A female with a single sex chromosome is affected with _____ syndrome.

2. Write short notes on the following (any 3) (3x6=18)

   (i) Dynamic mutations and anticipation

   (ii) PKU

   (iii) Genetic counselling

   (iv) Salient features of Human genome

3. (a) What is autosomal dominant inheritance? How is it different from autosomal recessive inheritance? Explain using hypothetical pedigree for each of the cases.

   (b) What are genetic variations? Explain the significance of these variations. (9,9)

4. (a) What are reciprocal chromosomal translocations? Explain in detail with an example.