KENYATTA UNIVERSITY

UNIVERSITY EXAMINATIONS 2016/2017

SECOND SEMESTER EXAMINATION FOR THE DEGREE OF MASTER OF SCIENCE IN MEDICAL BIOCHEMISTRY

SBC 880: GENETICS AND DEVELOPMENT ASPECTS OF DISEASE

DATE: Tuesday, 16th May 2017 **TIME: 9.00a.m-12.00p.m**

INSTRUCTIONS

Section A: Answer ALL questions (5 marks each)

1. Using an illustration, explain how mutations cause genetic diseases.

2. Describe the following concepts associated with chromosomal abnormalities
   (i) Aneuploidy
   (ii) Mosaicism

3. With the aid of an illustration explain molecular basis of genetic disorders.

4. Describe the chromosomal etiology and characteristics of Klinefelter syndrome

5. Describe how classic and clinical variant galactosemia is diagnosed.

6. Describe the following concepts associated with neoplasia
   (i) Cell immortalisation and tumourigenesis
   (ii) Cell oncogenes

7. Discuss the role of cytokines in the pathogenesis of rheumatoid arthritis (RA)

8. Outline the general features of sex chromosomal disorders.

Section B: Answer any THREE Questions (20 marks each)

1. Describe the etiology, symptoms and management of the following chromosomal abnormalities.

(i) Downer's [10 marks]
(ii) Tuners` [10 marks]

2. With a diagram, describe prenatal developmental changes of hemoglobin.

3. With a diagram, describe the pathogenesis and management of the following conditions:

(i) Phenylketonuria [10 marks]
(ii) Galactosemia [10 marks]

4. Using insulin and phenylalanine hydroxylase deficiency as examples, explain the biochemical and genetic basis of inborn errors.