NATIONAL UNIVERISTY OF SCIENCE AND TECHNOLOGY

FACULTY OF MEDICINE

MEDICAL SCHOOL

BACHELOR OF MEDICINE AND BACHELOR OF SURGERY DEGREE PART 1 EXAMINATIONS

(MCM 1202) : HUMAN GENETICS

DATE : DECEMBER 2005

TIME : 3 HOURS

Instructions to Candidates

1) Answer all questions

SECTION A

1. Discuss the causes and consequences of aneuploidy in humans with emphasis on the clinical implications. (20)

2. Write an essay on point mutations. (20)

SECTION B

1. In a family of 3 boys and 3 girls, two boys had hemophilia phenotype.

   a) Did they inherit the condition from the mother or father or from both parents? (3)
   b) Is the boy without hemophilia likely to be carrying its genotype? (2)
   c) Do the girls have a higher probability of carrying the hemophilia genotype. (3)
   d) If one more girl was born into the family could she possibly be hemophilic? (3)

2. Describe how RFLP is carried out in cases of paternal disputes. (11)

3. Why is the use of blood groups not recommended to solve paternity disputes? (3)

4. In the case of consanguinity, why is it that a lot of defects are expressed? (4)

5. In the case of deletion of a whole chromosome, why does its homology fail to function properly? (3)

6. What is minisatelite DNA? (4)

7. What is Alu DNA? (4)

8. What is an intron? (2)

9. How do you carry out Ames test for mutagenicity? (5)

10. Briefly, explain gene therapy. (5)

11. Outline two methods for chromosome banding. (8)

END OF EXAMINATION