

NATIONAL UNIVERSITY OF IRELAND, GALWAY
OLLSCOIL NA HEIREANN, GAILLIMH

SPRING EXAMINATIONS 2000

Third Science

GENETICS

EXTERN EXAMINER Professor C Brown
INTERN EXAMINER Internal Examiners

TIME ALLOWED: 3 hours.

ANSWER **FIVE** QUESTIONS

(INDICATE CLEARLY THE QUESTIONS ANSWERED ON THE FIRST
PAGE OF YOUR ANSWER BOOK)

- (1) Write an essay on "DNA damage and mutations with reference to the mechanisms required for their repair".
- (2) Write an essay describing the composition of a typical eukaryotic genome.
- (3) Write an essay entitled "The polymerase chain reaction (PCR) and its application in the diagnosis of genetic disease".
- (4) Write an essay on malfunctions of the immune system. Include in your answer, diseases that are directly and indirectly due to inheritance characteristics.
- (5) Write notes on two of the following:
 - a. Prenatal diagnosis
 - b. FISH (Fluorescence *in situ* hybridization)
 - c. Genetic imprinting
 - d. Gene mapping by meiotic recombination analysis.

- continued -

- (6) Calculate the frequency of the allele A_i in each of the following populations:

Population 1; consisting of 490 AA, 420 AA_i and 90 A_iA_i individuals. (10% of marks).

Population 2; consisting of 128 AA, 384 AA_i and 288 A_iA_i individuals. (10% of marks).

If all the individuals of both populations are mixed together what will be the frequency of A_i in the mixture? What will be the frequency of AA_i heterozygotes ? (20% of marks).

If all the individuals in the mixture now mate at random, what will be the frequency of the three genotypes in the offspring generation? (60% of marks).

- (7) Write an essay on "Aneuploidy of sex chromosomes and autosomes in the liveborn".
- (8) Write notes on two of the following:
- Huntington's Disease and Genetic Anticipation
 - Cystic fibrosis
 - Haemoglobinopathies
 - Polygenic inheritance
 - Tay Sachs Disease: include in your answer a mention of the role of intron mutation in this inherited disease.

- Good Luck and Best Wishes-