UNIVERSITY COLLEGE LONDON

University of London

EXAMINATION FOR INTERNAL STUDENTS

For The Following Qualifications:-

M.Sc. *PG Dip*

M.Sc. Prenatal Genetics & Fetal Medicine: Human Genetics

COURSE CODE	:	**PGFM0001**
DATE	:	**02-MAY-06**
TIME	:	**10.00**
TIME ALLOWED	:	**3 Hours**

TURN OVER

MSc /Diploma in Prenatal Genetics and Fetal Medicine
Human Genetics
2nd May 2006

There are two sections to this exam.
The short answer section makes up 25% of the marks for this paper.
The essay section counts for 75% of the marks (25% each essay).

Section 1 - SHORT ANSWER SECTION

This section makes up 25% of the examination.

1. **Write short notes on ALL of the following**

    a) FISH analysis giving two examples of its use in human genetics
    b) PCR analysis giving two examples of its use in human genetics
    c) Imprinting
    d) Mitochrondrial genetics
    e) DNA repair
    f) Prion disease

Section 2 - ESSAY SECTION

Answer THREE essays from this section.
Each essay counts for 25% of the examination.

1. Discuss the various mechanisms by which numerical abnormalities can arise in humans and the different stages of development at which these errors occur.

2. Compare and contrast techniques (cytogenetic and molecular) used in prenatal diagnosis with those used in preimplantation genetic diagnosis.

3. Give a detailed account of the genetics of triplet repeat disorders paying particular attention to the correlation between genotype and phenotype.

4. Describe the genetics of two of the following (include aspects such as inheritance, genotype-phenotype correlations, mutations, prevalence, gene discovery, gene function, therapeutic choices, etc.)

    (a) Beta- thalassaemia
    (b) Cystic Fibrosis
    (c) Charcot Marie Tooth disease

5. Discuss the statement 'Cancer is a genetic disease'.

END OF PAPER