

**UNIVERSITY COLLEGE LONDON**

University of London

**EXAMINATION FOR INTERNAL STUDENTS**

For The Following Qualifications:–

*Grad Dip M.Sc.*

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

**COURSE CODE : PGFM0001**

**DATE : 04-MAY-04**

**TIME : 14.30**

**TIME ALLOWED : 3 Hours**

**MSc in Prenatal Genetics and Fetal Medicine**  
**Human Genetics**  
**4<sup>th</sup> May 2004**

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) Gene therapy
  - b) Aneuploidy
  - c) Fluorescent in situ hybridisation
  - d) Polymerase chain reaction
  - e) Positional mapping of Mendelian diseases
  - f) Multifactorial disorders

**Section 2 - ESSAY SECTION**

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

1. Describe different types of genetic mutation and change and outline appropriate methods to detect them.
2. Using named examples, discuss tumour suppressor genes in cancer.
3. Outline meiosis. Compare the outcome of non-disjunction at meiosis I with meiosis II. What happens in meiosis in carriers of a) balanced translocations and b) inversions.
4. Describe the genetic aspects of TWO of the following:
  - (a) Huntington's disease
  - (b) Cystic Fibrosis
  - (c) DMD
5. Discuss the reproductive options and ethical dilemmas faced by families with a baby affected by beta-thalassaemia.

**END OF PAPER**