

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 : 03-MAY-05

TIME : 14.30

TIME ALLOWED : 3 Hours

MSc /Diploma in Prenatal Genetics and Fetal Medicine
Human Genetics
3rd May 2005

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) Karyotyping
- b) Linkage analysis
- c) Mutation detection
- d) Trinucleotide repeats
- e) Imprinting
- f) Familial cancer

Section 2 - ESSAY SECTION

Answer **THREE** essays from this section.

Each essay counts for 25% of the examination.

- 1. Compare and contrast meiosis in males and females.
- 2. Describe different numerical and structural chromosomal abnormalities and outline how they may affect the phenotype and fertility of carriers.
- 3. What is PCR? Discuss the applications of this technique in human genetic research and/or diagnosis.
- 4. Describe the genetic aspects of **TWO** of the following:
 - (a) Inborn errors of metabolism
 - (b) X-linked disorders
 - (c) Haemoglobinopathies
- 5. Outline genetic screening for a heritable disorder and discuss the advantages, problems and ethical issues associated with being identified as a mutation carrier.

END OF PAPER