

UNIVERSITY COLLEGE LONDON

University of London

EXAMINATION FOR INTERNAL STUDENTS

For the following qualifications :-

M.Sc.

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

COURSE CODE : PGFM0001

DATE : 03-MAY-02

TIME : 10.00

TIME ALLOWED : 3 hours

02-N0236-3-30

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TURN OVER

MSc in Prenatal Genetics and Fetal Medicine
Human Genetics
3rd May 2002

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) genetic screening
- b) karyotyping
- c) genetic polymorphism
- d) imprinting
- e) FISH
- f) some benefits of the information resulting from the human genome project

Section 2 - ESSAY SECTION

Answer three essays from this section.

Each essay counts for 25% of the examination.

1. Explain how Robertsonian and reciprocal translocations lead to problems during meiosis and how they affect fertility.
2. What are the consequences of mutation in human genes?
3. Write an essay on our current understanding of the genetic basis of cancer.
4. Describe the genetic aspects of two of the following:
 - (a) cystic fibrosis
 - (b) the triplet repeat disorders
 - (c) Duchenne muscular dystrophy
5. Discuss how advances in the understanding of the genetic basis of the haemoglobinopathies has been applied to the management of the disease in high risk populations.

END OF PAPER