OCR
RECOGNISING ACHIEVEMENT

OXFORD CAMBRIDGE AND RSA EXAMINATIONS

Advanced GCE

HUMAN BIOLOGY 2867

Genetics, Homeostasis and Ageing

Friday **24 JUNE 2005** Afternoon 2 hours

Candidates answer on the question paper.
Additional materials:
Electronic calculator
Ruler (cm/mm)

Candidate Name	Centre Number	Candidate Number

TIME 2 hours

INSTRUCTIONS TO CANDIDATES

- Write your name in the space above.
- Write your Centre number and Candidate number in the boxes above.
- Answer **all** the questions.
- Write your answers, in blue or black ink, in the spaces provided on the question paper.
- Read each question carefully before starting your answer.

INFORMATION FOR CANDIDATES

- The number of marks is given in brackets [ ] at the end of each question or part question.
- You will be awarded marks for the quality of written communication where this is indicated in the question.
- You may use an electronic calculator.
- You are advised to show all the steps in any calculations.

FOR EXAMINER'S USE		
Qu.	**Max.**	**Mark**
1	**10**	
2	**17**	
3	**21**	
4	**19**	
5	**18**	
6	**15**	
7	**20**	
TOTAL	**120**	

This question paper consists of 24 printed pages.

Registered Charity Number: 1066969
[Turn over

Answer all the questions.

1 Bacteria produce restriction enzymes which make cuts in double stranded DNA. Restriction enzymes can be extracted from bacteria and used to cut sections of DNA to be used in genetic engineering.

Fig. 1.1 shows a fragment of DNA which can be cut by the restriction enzyme Hind III. Hind III cuts the sequence AAGCTT, making a cut between the adenine bases to form sticky ends.

A diagram has been removed due to third party copyright restrictions

Details:

A diagram showing a fragment of DNA

Fig. 1.1

(a) (i) Complete Fig. 1.1 by writing in the letters for the missing nucleotide bases. [1]

(ii) Draw a line on Fig. 1.1 to show exactly where Hind III cuts the DNA. [1]

(iii) Describe the main features of a target site for a restriction enzyme.

.....

.....

.....

.....[2]

(b) Explain why DNA is described as complementary and antiparallel.

.....

.....

.....

.....

.....

.....[3]

(c) Explain why stem cells are ideally suited as host cells for gene therapy.

.....

.....

.....

.....

.....[3]

[Total: 10]

2 **(a)** The ability to remove excess heat is an important limiting factor in extending the duration of exercise.

Fig. 2.1 shows the change in core body temperature during exercise.

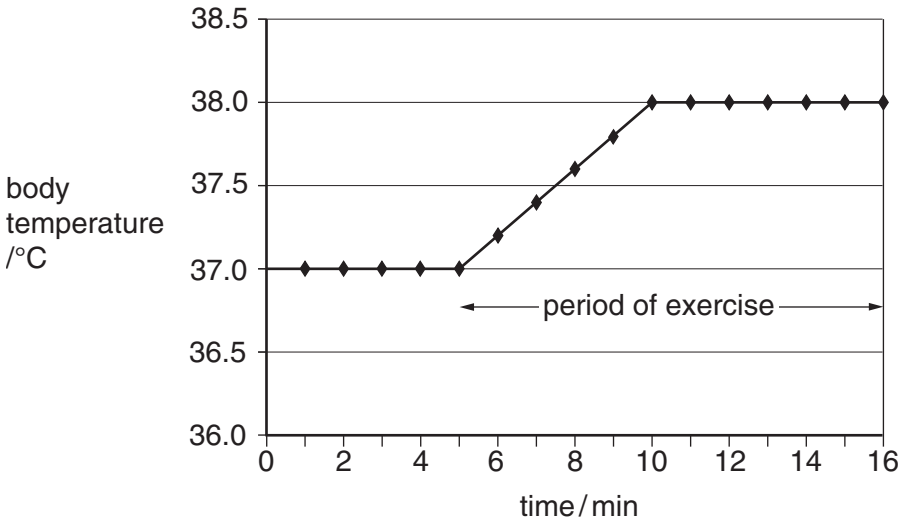

Fig. 2.1

(i) Describe how the body **normally** removes excess heat when the core temperature rises above 37 °C.

.....

.....

.....[2]

(ii) Explain why heat is produced during exercise.

.....

.....

.....

.....[2]

(b) Core body temperature is normally maintained at a set point (norm) of 37 °C.

(i) Why is it important to maintain the core temperature at a set point (norm) of 37 °C?

.....

.....

.....

.....

.....[3]

(ii) Suggest a reason for the slightly higher set point (norm) during exercise.

.....

.....

.....[1]

Question 2 continues on page 6.

(c)

- As environmental temperature varies, core body temperature must be controlled if the set point is to be maintained.
- Human populations originating in different temperature zones of the world have adapted to the temperature ranges found in these zones.
- Differences in mean body mass have evolved to make it easier to maintain the set point.

In an investigation, the mean body mass of a sample of the population in different temperature zones of the world was measured.

The results are shown in Fig. 2.2.

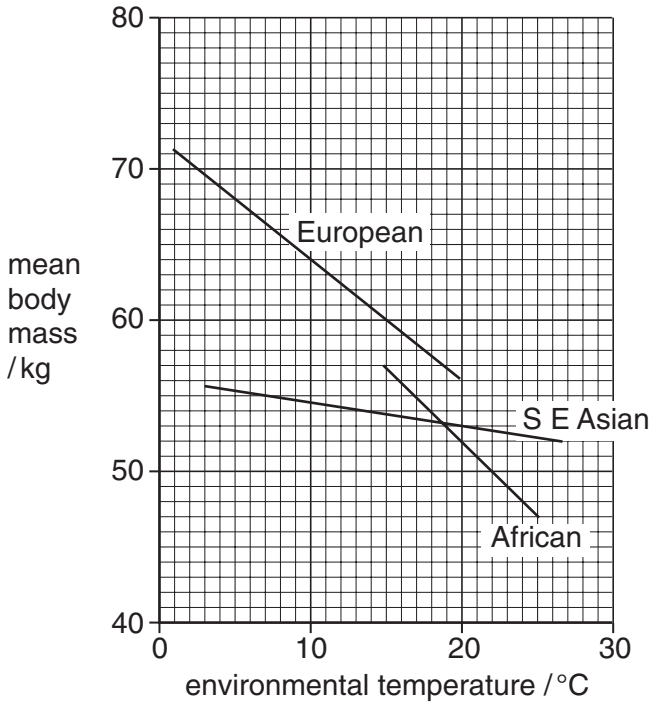

Fig. 2.2

(i) Calculate the percentage difference in the mean body mass of the European population at an environmental temperature of 5 °C compared with 15 °C. Show your working. **Give your answer to the nearest whole number**.

Answer = ...% [2]

(ii) Describe **and** explain the relationship between body mass and environmental temperature shown in the European population in Fig. 2.2.

.....

.....

.....

.....

.....

.....

.....[3]

(iii) Explain how the relationship between body mass and environmental temperature evolved in populations originating from different temperature zones.

.....

.....

.....

.....

.....[2]

(iv) Suggest **two other** environmental influences on body mass.

1 .....

.....

2 .....

.....[2]

[Total: 17]

3 (a) Analysis of the substances contained in a urine sample is useful in monitoring kidney function.

Fig. 3.1 shows a diagrammatic section of the glomerulus and Bowman's capsule in a nephron.

The effective filtration pressure (EFP) in the glomerulus depends on:

- the blood pressure in the glomerular capillaries (BP)
- the water potential of the plasma in the glomerular capillaries (WP)
- the pressure of the fluid in the Bowman's capsule (CP).

These pressures are shown by arrows on Fig. 3.1.

A diagram has been removed due to third party copyright restrictions

Details:

A diagram of a section of the glomerulus and Bowman's capsule in a nephron

Fig. 3.1

(i) Explain how each of the pressures shown in Fig. 3.1 is produced.

BP .....

.....

.....

WP .....

.....

.....

CP .....

.....

.....[3]

(ii) Write an equation, using the symbols given in **(a) (i)**, to show how these pressures interact to produce the EFP.

.....[1]

Question 3 continues on page 10.

(b) Table 3.1 shows the mean concentration of some of the substances in blood plasma, the glomerular filtrate and urine of an individual over 24 hours.

Table 3.1

	mean concentration / g dm^{-3}		
substances	plasma	glomerular filtrate	urine
protein	80.00	10.00	10.00
glucose	3.00	3.00	2.00
amino acids	0.50	0.50	0.00
urea	0.30	0.30	0.15

(i) Name the process which forms urine from the glomerular filtrate in the Bowman's capsule.

.....[1]

(ii) Table 3.1 shows an abnormally high concentration of protein and glucose in the urine.

Suggest an explanation for the abnormal concentrations of

protein .....

.....

.....

.....[2]

glucose .....

.....

.....

.....

.....

.....[4]

(c) In this question, one mark is available for the quality of use and organisation of scientific terms.

Explain how the volume of water excreted in urine is controlled.

.....

.....

.....

.....

.....

.....

.....

.....

.....

.....

.....

.....

.....

.....

.....

.....

.....

.....

.....[9]

Quality of Written Communication [1]

[Total: 21]

4 A rare genetic disease causes poor metabolism of lipid, and interferes with how lipid is stored. This results in symptoms which may affect many organs. The disease is caused by a single gene mutation, which will result in a specific enzyme deficiency. If both parents carry the same mutant allele, there is a one in four chance that their child will have the disease.

(a) (i) Choose **suitable** symbols to represent the normal and mutant alleles of this gene.

normal allele .....

mutant allele ..... [1]

(ii) Show by means of a genetic diagram how two phenotypically **normal** parents may produce a child who has this disease.

[3]

(iii) The disease occurs in equal numbers between males and females.

State the genetic terms used to describe the mutant allele.

.....[2]

(b) The symptoms resulting from the enzyme deficiency may involve many body organs, including the nervous system.

Explain why this enzyme deficiency

(i) may involve so many body organs;

.....

.....

.....[2]

(ii) may have a particularly severe effect on the nervous system.

.....

.....

.....[2]

(c) Some forms of this disease may be treated with long-term enzyme replacement therapy. The missing enzyme is injected at regular intervals throughout life and is successful as long as the injections begin soon after birth.

(i) State why the enzyme must be injected rather than taken as tablets.

.....

.....[1]

(ii) Suggest why the treatment must be started soon after birth to be successful.

.....

.....

.....[2]

(d) A small number of patients using enzyme replacement therapy develop antibodies in their blood to the injected enzyme.

(i) Explain why the release of antibodies is likely to make it difficult for patients to continue with this treatment.

.....

.....

.....

.....

.....

.....

.....

.....[4]

(ii) If antibodies develop, the dose in the enzyme injections is reduced or the injections are withdrawn.

Explain the risks involved in returning to the original dose of the enzyme after it has been reduced or withdrawn.

.....

.....

.....[2]

[Total: 19]

5 **(a)** One symptom of Alzheimer's disease is a reduction in the concentration of an enzyme in the brain that synthesises acetylcholine.

Outline the function of acetylcholine at cholinergic synapses.

.....

.....

.....

.....

.....

.....[4]

(b) Fig. 5.1 shows neurones from the brains of a healthy 70 year old, **A**, and a 70 year old with Alzheimer's disease, **B**.

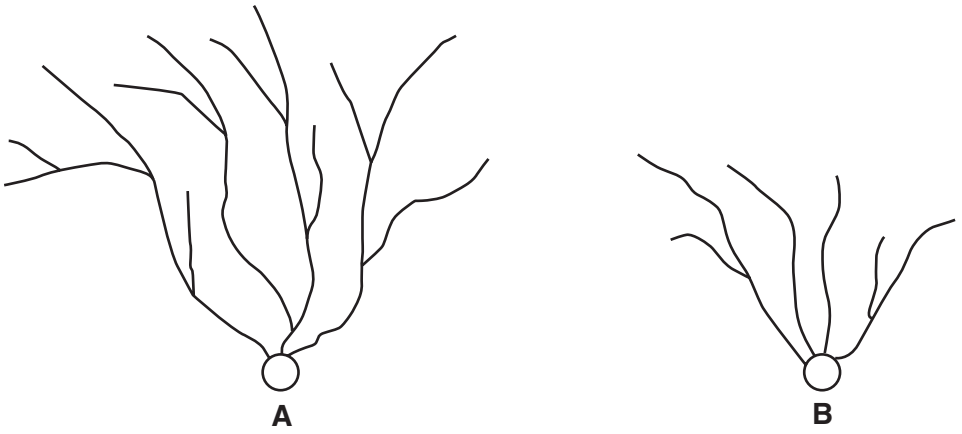

Fig. 5.1

(i) State **two** differences between the neurones shown in Fig. 5.1.

1 .....

.....

2 .....

.....[2]

(ii) Suggest how the differences in these neurones could account for the reduction in acetylcholine in a patient with Alzheimer's disease.

.....

.....[1]

(iii) Describe **one** recent development in treatment which promotes the regeneration of neurones.

.....

.....[1]

Question 5 continues on page 16.

(c) In this question, one mark is available for the quality of spelling, punctuation and grammar.

Fig. 5.2 is a diagrammatic section of the brain to show the areas most affected by the lack of acetylcholine.

A diagram has been removed due to third party copyright restrictions

Details:

A diagram of a section of the brain to show the areas most affected by the lack of acetylcholine

Fig. 5.2

Relate the symptoms of Alzheimer's disease to each of the areas of the brain shown in Fig. 5.2.

.....

.....

.....

.....

.....

.....

.....

.....

.....

.....

.....

.....

.....

.....

.....

.....

.....

.....

.....[9]

Quality of Written Communication [1]

[Total: 18]

6 **(a)** The risk of developing prostate cancer increases as a man ages.

(i) State the function of the prostate gland.

.....

.....[1]

(ii) Describe the signs and symptoms of prostate cancer **and** explain why it becomes more common as a man ages.

.....

.....

.....

.....

.....

.....

.....

.....

.....

.....[5]

(b) **(i)** The prostate specific antigen (PSA) test is the initial test for prostate cancer. PSA is a protein produced by the prostate gland, which is found in the blood in normal concentrations of 4 ng cm^{-3}. This concentration increases slightly as men age.

State what is meant by the term *prostate specific antigen*.

.....

.....[1]

(ii) If the prostate is enlarged by a benign or a cancerous growth, more PSA leaks into the blood. 60% of the men who have a PSA level above 10 ng cm^{-3} have prostate cancer.

Suggest why the PSA test is not available for a national screening programme for prostate cancer.

.....

.....

.....

.....

.....[3]

(iii) State **two** other techniques which could be used to detect prostate cancer.

1 .....

.....

2 .....

.....[2]

Question 6 continues on page 20.

(c) Fig. 6.1 shows the incidence per year of prostate cancer in different populations.

A graph has been removed due to third party copyright restrictions

Details:

A graph showing the incidence per year of prostate cancer in different populations

Fig. 6.1

Ageing has already been identified as a risk factor in the development of prostate cancer.

Describe what these data may suggest about other possible causes of prostate cancer. Support your answer using the information in Fig. 6.1.

.....

.....

.....

.....

.....[3]

[Total: 15]

7 All body cells, with the exception of erythrocytes, carry human leucocyte antigens (HLA). The antigens are coded for at four gene loci located close together on chromosome 6. The genes at these loci have many different alleles, which produce a huge variety of HLA haplotypes in the population.

Table 7.1 shows the number of possible alleles at each HLA locus.

Table 7.1

HLA locus	number of alleles
A	20
B	40
C	8
D	12

(a) How many HLA alleles are present in the genotype of an individual?

.....[1]

(b) How many haplotypes will a liver cell have?

.....[1]

(c) The number of possible different genotypes at each of the four loci is given by the formula

$$\frac{n\,(n+1)}{2}$$

where n is the number of possible alleles at that locus.

Using the information in Table 7.1 and the formula, calculate the number of possible genotypes at HLA locus **A**. Show your working.

Answer = ... [2]

(d) If a patient is to receive a successful liver transplant, the haplotypes of the donor and the recipient must match as closely as possible.

The probability of finding a close match from the general population is low.

State **two** reasons why, in practice, it may be easier to obtain a closer match from the general population than this statement suggests.

1 .....

.....

2 .....

.....[2]

(e) The haplotypes of a patient with liver failure are shown below.

A11	**A9**
B6	**B30**
C3	**C5**
D2	**D7**

The patient's partner, son and brother have all volunteered to be living donors.

Explain which individual is most likely to be a suitable donor.

.....

.....

.....

.....

.....

.....

.....[4]

(f) Healthy liver cells show an amazing capacity to form new liver tissue in a recipient, if rejection is avoided. Recently, three babies born with liver disease have been treated using healthy liver cells from a matching donor. The cells were injected into the babies' livers where they multiplied and functioned as normal liver cells.

(i) State **three** advantages of using injected liver cells rather than a transplanted liver.

1 .....

.....

2 .....

.....

3 .....

.....[3]

(ii) One of the babies had haemophilia, a condition in which the blood does not clot.

Explain the connection between liver disease in the baby and the failure of the blood to clot.

.....

.....

.....[2]

Question 7 continues on page 24.

(g) (i) Explain the genetic cause of haemophilia.

.....

.....

.....

.....[2]

(ii) Explain how haemophilia is inherited. You may use a genetic diagram if it makes your answer clearer.

.....

.....

.....

.....

[3]

[Total: 20]

END OF QUESTION PAPER