OCR
RECOGNISING ACHIEVEMENT

ADVANCED GCE **2867**

HUMAN BIOLOGY

Genetics, Homeostasis and Ageing

FRIDAY 22 JUNE 2007 Afternoon

Time: 2 hours

Additional materials: Electronic calculator
Ruler (cm/mm)

CUP/T19809

Candidate Name

Centre Number

Candidate Number

INSTRUCTIONS TO CANDIDATES

- Write your name, Centre Number and Candidate Number in the boxes above.
- Answer **all** the questions.
- Use blue or black ink. Pencil may be used for graphs and diagrams only.
- Read each question carefully and make sure you know what you have to do before starting your answer.
- Do **not** write in the bar code.
- Do **not** write outside the box bordering each page.
- WRITE YOUR ANSWER TO EACH QUESTION IN THE SPACE PROVIDED. ANSWERS WRITTEN ELSEWHERE WILL NOT BE MARKED.

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	16	
2	19	
3	16	
4	18	
5	19	
6	17	
7	15	
TOTAL	120	

This document consists of **21** printed pages and **3** blank pages.

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Answer **all** the questions.

1 Maintaining the body temperature at a set point (norm) is essential if the body is to function efficiently.

(a) (i) State the set point for body temperature.

.....[1]

(ii) Name the part of the **central nervous system** and the part of the **peripheral nervous system** that maintains the body temperature at its set point.

central .....

peripheral .....[2]

(iii) Outline how body temperature is kept at its set point.

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(b) The body size and shape of humans in an ethnic population are adaptations closely related to the environmental temperature.

The human populations with the lowest mass in proportion to their height are Nilotic populations from the desert areas of North Africa. Those with the greatest mass in proportion to their height are the Inuit populations from the Arctic and Northern Canada.

Fig. 1.1 shows a Nilotic and an Inuit male of similar ages.

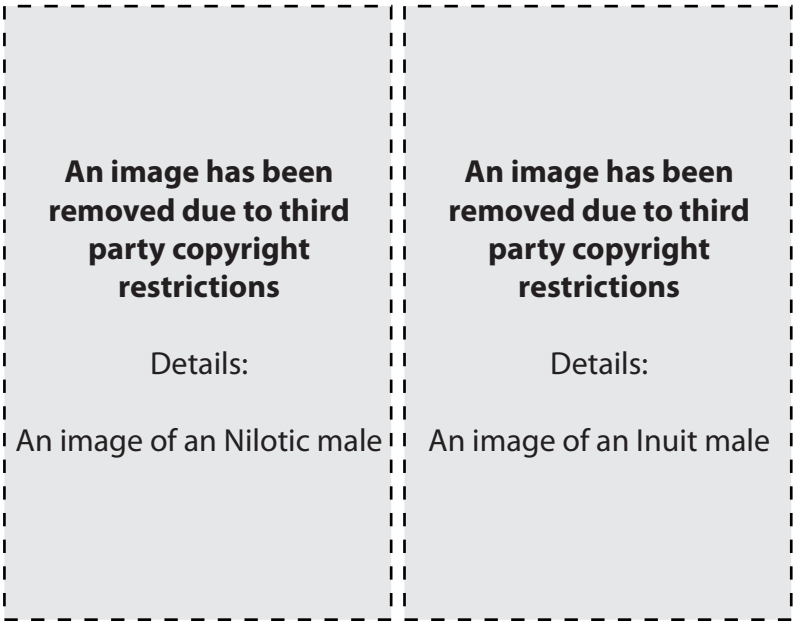

Fig. 1.1

Table 1.1 shows the mass and height of the two men.

Table 1.1

	Nilotic	Inuit
mass/kg	68	68
height/m	1.90	1.60

The ratio of mass to height is called the Body Mass Index (BMI). It is calculated using the formula:

$$\frac{\text{body mass}}{\text{height}^2}$$

(i) The BMI of the Nilotic male is 18.84.

Using the information in Table 1.1, calculate the BMI of the Inuit male.

Show your working. Give your answer to two decimal places.

Answer = ... [2]

(ii) The Nilotic man is adapted for life in the hot deserts of North Africa and the Inuit man for life in the frozen areas of the Arctic.

Using the information in Fig. 1.1 and Table 1.1, explain the differences in the BMI of the Nilotic and Inuit men **and** suggest how these differences adapt them for life in their environment.

Nilotic male .....

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Inuit male .....

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[Total: 16]

2 **(a)** Abnormalities of the immune system are responsible for a large number of disease conditions.

It has been discovered that some people have antibodies in their blood which complement the cell surface antigens on beta cells in their pancreas.

Suggest what is unusual about these antibodies **and** describe their effects on the beta cells in the pancreas.

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(b) The normal concentration of glucose in the blood is 90 – 100 mg per 100 cm^3 of blood.

Table 2.1 shows the results of glucose tolerance tests on two individuals, **A** and **B**.

- Their blood glucose concentration was measured at the start of the test.
- They were then given 50 g of glucose.
- Their blood glucose concentration was then measured at fifteen-minute intervals for two hours.

Table 2.1

time / min	blood glucose concentration / mg per 100 cm^3 blood	
	A	**B**
0	90	100
15	110	125
30	140	172
45	100	190
60	80	208
75	91	205
90	84	200
105	87	197
120	89	195

(i) Explain why the blood glucose concentration of individual **A** did not return immediately to 90–100 mg per 100 cm^3 blood.

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(ii) Explain the data for individual **B** given in Table 2.1.

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(c) Individuals with Type 2 diabetes are recommended to make the following changes to their diet:

- avoid foods high in refined sugar (a high glycaemic index)
- eat carbohydrate in the form of more starch (a low glycaemic index)
- reduce the intake of saturated fat.

(i) Explain why it is important to avoid foods high in refined sugar but to eat more starch.

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(ii) Suggest why diabetics should eat less saturated fat.

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(d) Diabetics may also be prescribed statins. These are drugs that lower blood cholesterol.

In a clinical study, patients with Type 2 diabetes were divided into three groups:

- group **X** was treated by changes in the diet only
- group **Y** was treated by statin only
- group **Z** was treated by both diet and statin.

The following measurements were taken from each group:

- blood LDL concentration
- blood insulin concentration
- resistance to the effect of insulin.

After 12 weeks, the three measurements were repeated and the **mean percentage change** calculated for each group.

The results of this investigation are shown in Fig. 2.1.

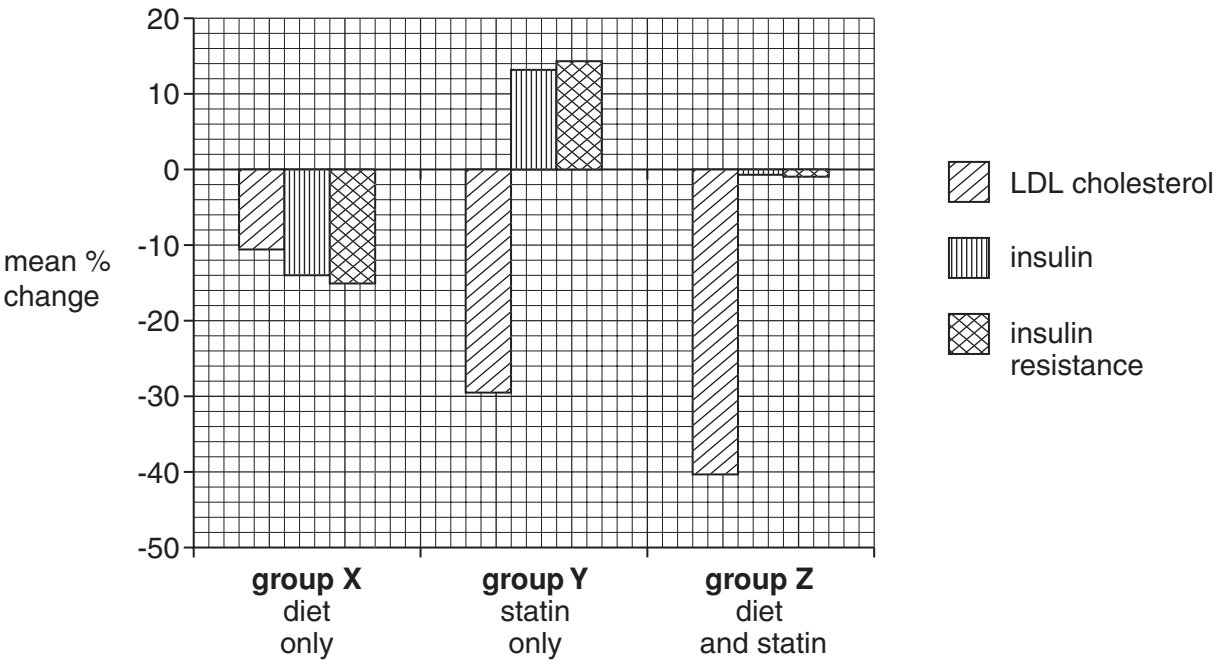

Fig. 2.1

Using the information in Fig. 2.1, explain why diabetics are encouraged to use a combination of diet and statin, rather than diet or statin alone, to treat their condition.

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[Total: 19]

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

An elderly gentleman asks his daughter if she will take him to a nearby department store to buy a new telephone, as he is experiencing difficulty with the telephone he has at present.

- He cannot easily hear the caller when he answers the phone.
- When he makes a call he frequently dials the wrong number.
- He also has difficulty finding the right buttons to press.

(a) Describe the changes that may have occurred in the **brain** and **sense organs** as the gentleman ages, which could account for his difficulty with the telephone.

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Quality of Written Communication [1]

(b) The daughter is worried that this is not the first time that her father has asked her for help with routine tasks. She suspects that he may be developing a degree of dementia.

(i) Describe **one** suitable diagnostic procedure which would help to assess her father's condition.

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(ii) Outline the **long term** difficulties that may be faced by the **daughter** if her father is found to have dementia.

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(iii) Suggest **two** ways in which the daughter could seek extra help for her father.

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[Total: 16]

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

Heart transplants may be recommended for severe coronary heart disease, heart failure or for infants born with severe heart defects.

Fig. 4.1 shows where a transplanted heart is attached inside the body.

A diagram has been removed due to third party copyright restrictions

Details:

A diagram showing where a transplanted heart is attached inside the body

Fig. 4.1

Outline the practical and ethical issues involved in transplant surgery.

practical issues .....

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ethical issues .....

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Quality of Written Communication [1]

(b) A patient was given a heart transplant for severe coronary heart disease (CHD).

Suggest the advice that might be given to this patient for his after-care.

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(c) A new technique has been developed in the United States that allows the transplant of a kidney from a living donor who does **not** have a matching blood group.

The technique involves 'washing' the blood of the recipient three times a week for three months, to remove all the antibodies that would damage the transplanted kidney.

(i) A patient who is blood group **O** has been offered a kidney transplant from a relative who is group **AB**.

State the **antibodies** from the **ABO** system that must be 'washed' out of the blood if the transplant is to be successful.

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(ii) In addition to the blood groups, it is important that the tissue type should match as closely as possible.

Explain what is meant by the *tissue type* in this context.

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[Total: 18]

5 The gene that causes nail patella syndrome is linked to the **ABO** blood group gene. This is one of the few examples of autosomal linkage in humans that is well understood.

(a) The Human Genome Project has revealed other autosomal linkage groups. This has contributed greatly to the mapping of human chromosomes and has considerable potential for the diagnosis of human genetic diseases.

Suggest how linked genes may have the potential for the diagnosis of genetic diseases.

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As known examples of autosomal linkage in humans are very rare, an imaginary example has been used in the following questions.

It has been proposed that individuals who have a long index finger (locus **D**/**d**) also have a short big toe (locus **T**/**t**).

An individual with a long index finger and a short big toe has five children with a partner who has a normal length index finger and a normal length big toe.

All their children have a normal length index finger and a normal length big toe.

(b) State whether the allele for a long index finger is dominant or recessive. Give a reason for your answer.

allele .....

reason .....

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.....[2]

(c) Using the symbols **D**/**d** for the index finger length locus and **T**/**t** for the toe length locus,

(i) state the genotype of the children;

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(ii) state the possible genotypes of the parent with a normal length index finger and a normal length big toe.

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(d) Researchers investigated a large sample of people with the heterozygous genotype for both index finger and big toe length. Each person had children with a partner who had a long index finger and a short big toe.

The number of children with each phenotype is shown below.

- long index finger and a short big toe 83
- long index finger and a normal big toe 21
- normal index finger and a short big toe 20
- normal index finger and a normal big toe 80

The ratio of phenotypes for unlinked loci is predicted to be 1 : 1 : 1 : 1

The chi-squared (χ^2) test was conducted on these data, giving a value for χ^2 of 73.06.

Table 5.1

degrees of freedom	probability (p)				
	0.10	0.05	0.02	0.01	0.001
1	2.71	3.84	5.41	6.64	10.83
2	4.61	5.99	7.82	9.21	13.82
3	6.25	7.82	9.84	11.35	16.27
4	7.78	9.49	11.67	13.28	18.47

(i) Use the value of χ^2 and Table 5.1 to find the probability of these results differing from the expected ratio by chance.

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(ii) State the conclusions that may be drawn from the probability found in **(d)(i)**.

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(iii) The difference between the observed and expected results in this investigation occurs because the loci for index finger length and big toe length are on the same chromosome (linked).

Explain **by means of a genetic diagram**, how the actual results of these crosses occurred.

parental phenotypes	normal index finger normal big toe	long index finger short big toe
parental genotypes	.....	.....
gametes	.....	.....

explanation .....

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[Total: 19]

6 Scientists have discovered that cells scraped from the germinal epithelium at the surface of the ovary can develop into mature human eggs (secondary oocytes) and granulosa cells.

The cells removed from the germinal epithelium are originally pluripotent. These cells only form secondary oocytes and granulosa cells when placed in a suitable culture medium.

Fig. 6.1 is a light microscope photograph of a transverse section through the ovary.

Fig. 6.1

(a) (i) Explain what is meant by the phrase *a suitable culture medium* in this context.

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(ii) Pluripotent cells are able to develop into many types of cell if they are cultured in a suitable medium.

State one *other source* of pluripotent human cells.

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(iii) Suggest the risks of using germinal epithelial cells from an older woman to produce secondary oocytes.

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(b) It has been suggested that secondary oocytes could be used to regain fertility in menopausal women.

(i) Suggest how this might delay the menopause.

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(ii) Discuss the **advantages** and **disadvantages** of delaying the menopause.

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(c) Discuss methods, **other than the use of secondary oocytes**, that may be used to delay the menopause or to reduce its symptoms.

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(d) Secondary oocytes produced from germinal epithelium could become a valuable resource for IVF.

Explain how IVF could change the frequency in the population of alleles that cause disease.

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[Total: 17]

7 As the diagnosis of genetic disease improves, there is an increased need for counselling individuals who are involved.

(a) Turner's syndrome is caused by a chromosomal mutation.

(i) Suggest why this condition is called a *syndrome*.

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(ii) Describe how Turner's syndrome occurs.

You may use a labelled diagram if it helps you to answer the question.

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(b) Turner's syndrome may be diagnosed while the foetus is still in the uterus, by obtaining cells from the amniotic fluid.

(i) Outline how these cells are treated to diagnose Turner's syndrome.

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(ii) Why are chromosomal mutations easier to diagnose than gene mutations?

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(c) The improved diagnosis of genetic disease involves some important ethical issues.

Describe two **ethical issues** that may be involved.

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[Total: 15]

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