OCR
RECOGNISING ACHIEVEMENT

ADVANCED GCE **2867**

HUMAN BIOLOGY

Genetics, Homeostasis and Ageing

WEDNESDAY 30 JANUARY 2008 Afternoon

Time: 2 hours

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

CUP/T57250

Candidate Forename		Candidate Surname	

Centre Number						Candidate Number				

INSTRUCTIONS TO CANDIDATES

- Write your name in capital letters, your Centre Number and Candidate Number in the boxes above.
- Use blue or black ink. Pencil may be used for graphs and diagrams only.
- Read each question carefully and make sure that you know what you have to do before starting your answer.
- Answer **all** the questions.
- Do **not** write in the bar codes.
- Do **not** write outside the box bordering each page.
- Write your answer to each question in the space provided.

INFORMATION FOR CANDIDATES

- The number of marks for each question is given in brackets [ ] at the end of each question or part question.
- The total number of marks for this paper is **120**.
- 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	**14**	
2	**19**	
3	**15**	
4	**19**	
5	**17**	
6	**18**	
7	**18**	
TOTAL	**120**	

This document consists of **20** printed pages and **4** blank pages.

[Turn over

Answer **all** the questions.

1 The origin of *Homo sapiens sapiens* is a topic which has fascinated generations of scientists.

How did we evolve? Are we still evolving? Why are there no other *Homo* species alive today?

(a) The study of the evolutionary ancestors of *H. sapiens sapiens* involves the science of taxonomy.

Explain the meaning of the term *taxonomy*.

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..... [3]

(b) Table 1.1 shows the main taxonomic groups.

Table 1.1

	P	Q	R	S	T	U	V
taxonomic group	species	order	family	kingdom	class	phylum	genus

Place the letters representing the taxonomic groups in the correct order.

The first one has been done for you.

S .... [3]

(c) Suggest why *H. sapiens sapiens* is considered by some taxonomists to be a **subspecies**.

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..... [1]

(d) Explain how geographical isolation can lead to genetic isolation in the development of new species.

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(e) Explain why there has been no further speciation of *H. sapiens sapiens*.

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(f) Different species cannot breed together to produce fertile young.

Explain why they cannot produce fertile young.

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..... [1]

[Total: 14]

2 Research into the causes of genetic diseases is developing fast. Many of these diseases are caused by mutations.

(a) **(i)** Define the term *mutation*.

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..... [2]

(ii) A mutation in a somatic cell, such as a skin cell, may not be as serious as a mutation in a germ cell, such as a primary oocyte.

Suggest why this is so.

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(b) DNA is present in the nuclei of cells. Cells also contain DNA in the matrix of their mitochondria.

Mitochondrial DNA (mtDNA) has been used to provide evidence of human evolution and how closely related different human groups are.

(i) DNA from the nucleus is passed on from one generation to the next by the male and female gametes. mtDNA, however, is only passed on by the female gamete.

Suggest why mtDNA is only passed on by the female gamete.

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..... [1]

(ii) Suggest why mtDNA is so useful in providing evidence of human evolution **and** evidence of how closely related human groups are.

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..... [2]

(c) Mutations in mtDNA can cause genetic diseases, although these are rare.

Suggest why genetic diseases caused by mtDNA are rare.

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..... [1]

(d) Leber Hereditary Optic Neuropathy (LHON) is a rare disease which causes an affected individual to suffer an acute loss of vision. This loss of vision is permanent.

One of three possible point mutations in the mtDNA causes the disease to develop.

The prevalence of each mutation varies in different ethnic groups.

(i) Explain the meaning of the following terms in this context.

term	meaning
acute	
prevalence	
point mutation	

[3]

(ii) A woman who has a mother with LHON is hoping to start a family and so consults a genetic counsellor.

Outline the information that a genetic counsellor might give to the woman.

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..... [4]

(e) Research on LHON has shown that in most cases the blindness develops because the optic nerve has stopped functioning.

Mutations in mtDNA usually disrupt the metabolic pathway for aerobic respiration.

Suggest why this mutation may cause the optic nerve to stop functioning.

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..... [3]

[Total: 19]

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3 The government of the United Kingdom is becoming increasingly concerned by the expected rapid rise in the elderly members of the population.

The concern is that there will be relatively more elderly people compared with those who are younger and more likely to be contributing to the economy.

(a) In Table 3.1, the population over 65 is shown as a percentage of those who are aged between 15 and 64.

These data have been projected to 2050.

Table 3.1

year	population over 65 as a % of population aged between 15 and 64
2005	24
2010	25
2015	28
2020	30
2025	33
2030	37
2035	39
2040	40
2045	39
2050	39

(i) Describe the trend shown by the data in Table 3.1.

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..... [2]

(ii) Suggest **two** reasons for this trend.

1 .....

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2 .....

..... [2]

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

Discuss the advantages **and** disadvantages of an ageing population to society.

Credit will be given if you include the information in Table 3.1 in your answer.

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Quality of Written Communication [1]

(c) Elderly people may be more at risk from infection.

(i) Explain why this is so.

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..... [2]

(ii) State **one** precaution that should be taken by elderly people to reduce the risk of infection.

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..... [1]

[Total: 15]

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4 Do you feel healthy and 'in balance'? If the answer is yes, your autonomic nervous system (ANS) is working efficiently. It is controlling the homeostatic mechanisms in your body and maintaining the dynamic equilibrium of your body constants.

Many homeostatic mechanisms are regulated by the hypothalamus and the pituitary gland.

Fig. 4.1 is a diagram of the hypothalamus and the pituitary gland.

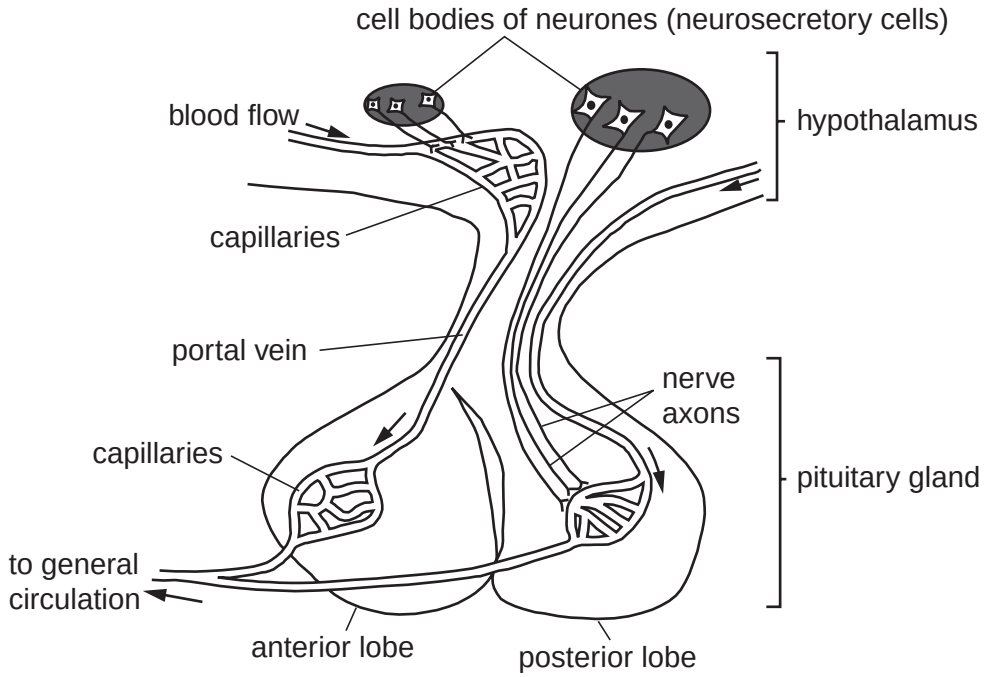

Fig. 4.1

(a) (i) Select the correct location for the hypothalamus **and** pituitary gland from the list below, by placing a tick in the correct box.

below the cerebellum	
below the forebrain	
below the midbrain	
below the hindbrain	

[1]

(ii) State **two** homeostatic mechanisms that are controlled by the hypothalamus.

1 .....

2 ..... [2]

(iii) Using Fig. 4.1, describe why the portal vein between the hypothalamus and the anterior lobe of the pituitary gland is unusual when compared with most veins in the body.

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..... [1]

(iv) Using Fig. 4.1, suggest the specialised function of the neurosecretory cells in the hypothalamus.

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..... [2]

(v) Complete Table 4.1 to show the hormones, their functions and whether they are produced in the hypothalamus or in part of the pituitary gland.

Table 4.1

hormone	function	where produced
thyroid stimulating hormone (TSH)		anterior pituitary gland
	stimulates contraction of uterine muscle	hypothalamus
follicle stimulating hormone (FSH)		anterior pituitary gland
	controls water balance by increasing permeability of collecting ducts	hypothalamus

[4]

(b) Children are checked regularly at health centre clinics to make sure that their growth and development fall within normal limits.

It was noticed at a clinic that one child was extremely tall for their age. The child's height was several standard deviations above the mean for children of the same sex and age in the population.

If this condition continues past puberty it is called gigantism. Gigantism is caused by an excess of growth hormone.

(i) Explain what is meant by the term *standard deviation*.

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..... [2]

(ii) Further investigation revealed that the child had a slow-growing benign tumour in the anterior pituitary gland.

Suggest how this tumour could cause gigantism.

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..... [3]

(iii) Although the tumour was benign, it was decided that treatment was necessary.

Suggest **two** reasons why it was decided that treatment was necessary.

1 .....

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2 .....

..... [2]

(iv) State a suitable treatment for this tumour **and** give a reason for your choice.

treatment .....

reason .....

..... [2]

[Total: 19]

5 The kidneys are one of the most important organs in the body and their functions are essential for survival. Each kidney receives 1200 cm^3 of blood per minute from the left ventricle.

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

Describe the **gross** structure of the kidney **and** explain the importance of the kidney in the normal functioning of the body.

Details of how the kidney performs these functions are **not** required.

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Quality of Written Communication [1]

(b) Cancer of the kidney can occur. The cause is not known but there is some evidence to suggest that cigarette smoking increases the risk.

The outcome of the disease varies depending on whether cells from the cancer have broken away. Early diagnosis is essential if treatment is to be successful.

(i) Suggest how smoking may increase the risk of kidney cancer.

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..... [2]

(ii) Explain how cells breaking away from the cancer affects the outcome of the disease.

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..... [3]

(iii) Suggest an **early warning** sign that a patient might spot for kidney cancer.

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..... [1]

(iv) Describe a technique which could be used to **confirm** the diagnosis of kidney cancer.

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..... [2]

[Total: 17]

6 Mary is 16 years old and lately has been feeling unwell. She went to the doctor and was told that she had Type 2 diabetes. Mary thought that Type 2 diabetes was an illness of older people.

Mary often went out with her friends and liked nothing better than to round off the evening with a burger and chips and a bottle of fizzy drink. She has been gaining too much weight over the last few years.

The doctor gave Mary a glucose testing meter to test her blood glucose concentration every morning before breakfast.

(a) Explain why the doctor wanted Mary to:

(i) measure her blood glucose concentration;

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(ii) measure it every morning before breakfast.

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[5]

(b) The doctor explained to Mary that it was essential that she changed her diet.

State the changes that Mary should make to her diet.

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..... [3]

(c) Gross proteinuria is a condition where large quantities of protein appear in the urine. Research has shown that if diabetes is not treated and blood glucose controlled, proteinuria may develop within 10 years of diagnosis of diabetes.

Gross proteinuria is an indicator of severe kidney damage, which may eventually develop into renal failure.

Fig. 6.1 shows the incidence of diabetes and the incidence of gross proteinuria, 10 years after diagnosis of diabetes.

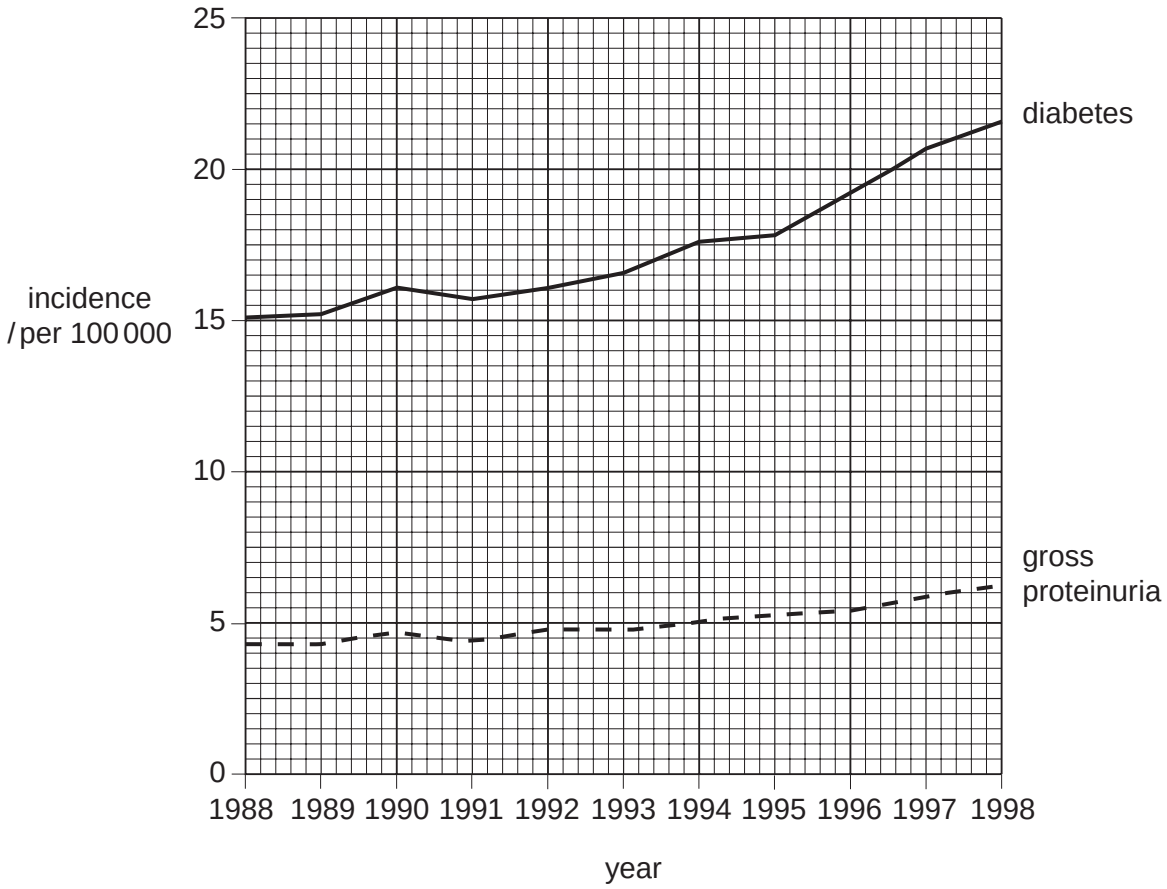

Fig. 6.1

(i) Calculate the percentage increase in the incidence of **diabetes** from 1988 to 1998.

Show your working and give your answer **to one significant figure**.

Answer = ... % [2]

(ii) Describe the relationship between the incidence of diabetes and gross proteinuria, as shown in Fig. 6.1.

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(d) Gross proteinuria is usually accompanied by excessively high blood pressure (hypertension) in the capillaries of the glomeruli. This damages the basement membrane in the endothelium of the capillaries.

(i) Describe the function of the basement membrane in the endothelium of **normal** capillaries in the glomerulus.

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(ii) Describe the likely effect of hypertension in the capillaries of the glomerulus.

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[Total: 18]

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7 There are a number of diseases where the immune system does not function as well as it should. These immunodeficiency diseases are either **inherited** or **acquired** during an individual's lifetime.

(a) Explain the difference between these two ways of becoming 'immunodeficient'.

inherited .....

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acquired .....

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..... [3]

(b) One inherited type of immunodeficiency disease causes an absence of B lymphocytes and plasma cells.

(i) Explain the connection between B lymphocytes and plasma cells.

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..... [2]

(ii) State the type of organism that would **not** be destroyed by an immune system without B lymphocytes.

..... [1]

(iii) This immunodeficiency disease does not produce any obvious symptoms and therefore is rarely diagnosed until the individual is between two months and two years of age.

Suggest why symptoms may not occur **until this age**.

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..... [2]

(c) The immunodeficiency disease described in **(b)** is a sex-linked recessive condition.

(i) Choose a suitable symbol to represent the recessive allele that may be present at the gene locus.

..... [1]

(ii) Explain how this disease is inherited.

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..... [3]

(iii) Suggest how this disease could be treated.

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(d) Severe Combined Immune Deficiency (SCID) is a severe form of inherited immunodeficiency disease where both B **and** T lymphocytes do not form.

Symptoms develop during the first few months of life and will result in death during the first two years, unless the infant is kept in a sterile environment.

Discuss the **ethical** issues affecting the families in which severe inherited diseases occur.

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[Total: 18]

END OF QUESTION PAPER

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