

**ADVANCED GCE****HUMAN BIOLOGY**

Genetics, Homeostasis and Ageing

2867

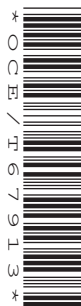
Candidates answer on the question paper

OCR Supplied Materials:

None

Other Materials Required:

- Electronic calculator
- Ruler (cm/mm)

Wednesday 17 June 2009**Afternoon****Duration: 2 hours**

Candidate Forename		Candidate Surname	
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Centre Number						Candidate Number				
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INSTRUCTIONS TO CANDIDATES

- Write your name clearly in capital letters, your Centre Number and Candidate Number in the boxes above.
- Use 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.
- Write your answer to each question in the space provided, however additional paper may be used if necessary.

INFORMATION FOR CANDIDATES

- The number of marks 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.
- This document consists of **24** pages. Any blank pages are indicated.

FOR EXAMINER'S USE

Qu.	Max.	Mark
1	15	
2	14	
3	15	
4	17	
5	19	
6	22	
7	18	
TOTAL	120	

Answer **all** the questions.

- 1 Investigating changes in the composition of urine is a quick and easy means of detecting abnormalities in body function. Urine contains a number of excretory products.

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

.....
 [1]

(ii) Name **two** excretory products that are found in the urine of a healthy person.

1
 2 [2]

(iii) Carbon dioxide is an excretory product that is **not** found in urine.

Explain why it is important to remove carbon dioxide from the body.

.....

 [3]

(b) State **two** changes in the composition of urine that may indicate a disease condition.

Explain what has caused each of the changes in the composition of urine.

change 1
explanation

.....

change 2
explanation

.....
 [6]

- (c) In people with kidney or bladder cancer, the urine contains by-products of the disease.

An investigation was conducted to see if it is possible to train dogs to detect the by-products of the disease by smelling samples of human urine.

The investigation was also carried out to find out if this technique could be used as a diagnostic test.

- (i) State what is meant by a 'diagnostic test' in this context.

.....
 [1]

- (ii) The dogs identified the cancer sample correctly on 22 out of 54 occasions.

The expected success rate due to chance was 14%. The dogs achieved a success rate of 41%.

The test is not yet used routinely for screening for kidney and bladder cancer.

Suggest the ethical problems involved in using this method for the diagnosis of kidney and bladder cancers.

.....

 [2]

[Total: 15]

- 2 If the body is to remain healthy it is important that it is able to detect and respond to changes in its internal and external environments.

This function is carried out by the nervous and endocrine systems.

- (a) Explain what is meant by an endocrine gland.

.....

.....

..... [2]

- (b) Complete the table below by stating **three** differences between the ways in which the nervous and endocrine systems work.

nervous system	endocrine system

[3]

- (c) State what is meant by the 'internal environment'.

.....

..... [1]

- (d) The nervous and endocrine systems can respond to both internal and external stimuli.

Name **one** internal stimulus and **one** external stimulus that each system can respond to.

- (i) Nervous system:

internal stimulus

external stimulus [2]

- (ii) Endocrine system:

internal stimulus

external stimulus [2]

- (e) Fig. 2.1 shows the normal effect on the concentration of blood glucose of taking small, regular doses of glucose by mouth.

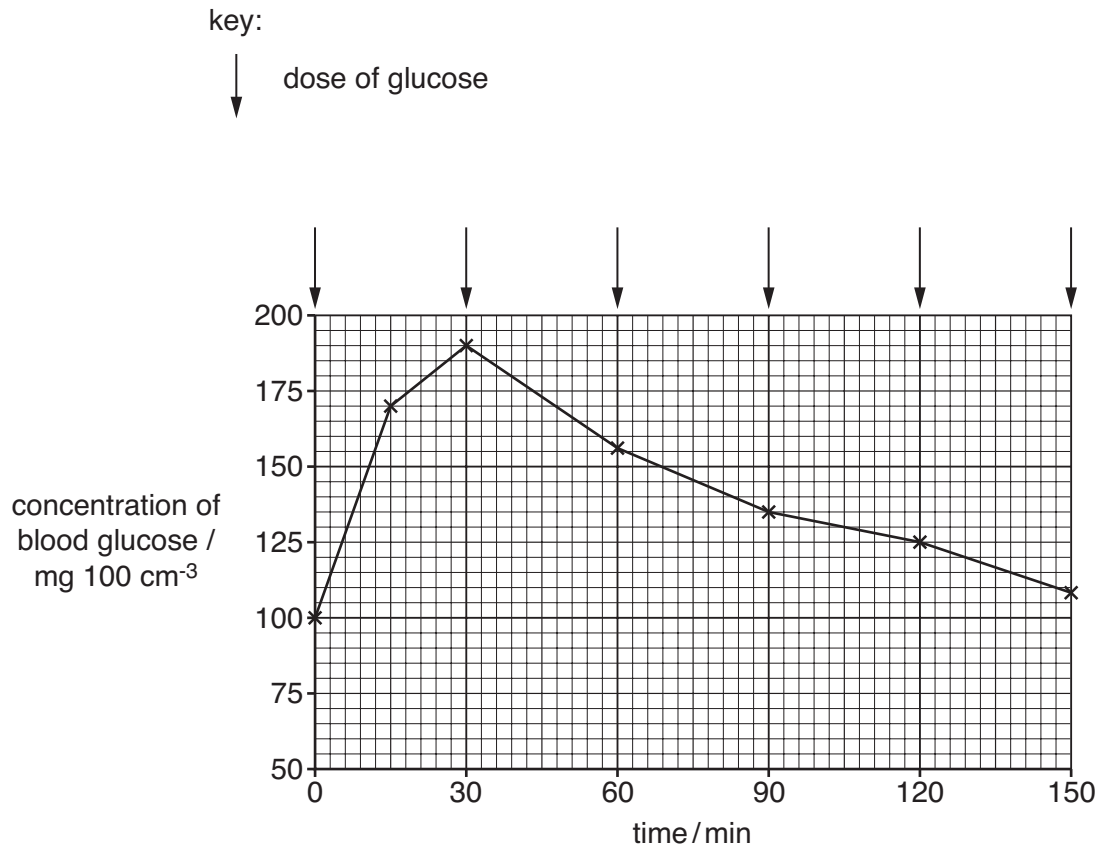


Fig. 2.1

Explain why the last three doses of glucose do **not** have the same effect as the first dose.

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..... [4]

[Total: 14]

Turn over

- 3 Down's syndrome is a genetic condition that may delay physical, intellectual and language development. It is usually caused by the failure of chromosome 21 to separate during gamete formation in the mother.

(a) (i) State the correct term for the failure of chromosome pairs to separate.

..... [1]

- (ii) Explain why an individual with this form of Down's syndrome will have **three** copies of chromosome 21 (trisomy 21) as a result of the failure of pair 21 to separate during gamete formation in the mother.

You may illustrate your answer with a diagram.

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..... [2]

- (b) Table 3.1 shows the relationship between the age of the mother and the incidence of Down's syndrome per two hundred live births.

Table 3.1

age of mother / years	incidence of Down's syndrome per 200 live births
15–19	0.10
20–24	0.18
25–29	0.20
30–34	0.30
35–39	0.80
40–44	2.40
45+	6.40

- (i) Calculate the percentage increase in the risk of having a child with this form of Down's syndrome between the ages of 20–24 and 35–39.

Show your working and give your answer to **two decimal places**.

Answer =% [2]

- (ii) Using the information in Table 3.1, describe the relationship between Down's syndrome and the age of the mother.

.....

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..... [2]

- (iii) There is no relationship between the age of the father and the incidence of Down's syndrome.

Suggest why the relationship between age and incidence exists in the mother **and** not in the father.

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..... [3]

- (c) Trisomy 21 is not the only cause of Down's syndrome.

Describe **and** explain another type of chromosomal mutation that can cause Down's syndrome.

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..... [3]

- (d) Screening for Down's syndrome is a routine part of antenatal care. This is now offered to most pregnant women regardless of age.

The first tests offered to the mother are a blood test and an ultrasound scan. If, in light of the results of these tests, Down's syndrome is suspected, amniocentesis or chorionic villus sampling (CVS) is advised.

Suggest why the first screening tests offered to pregnant women are blood tests and ultrasound scans rather than amniocentesis or CVS.

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..... [2]

[Total: 15]

- 4 The thyroid gland absorbs iodine ions from the blood to make thyroxine. To diagnose disorders of the thyroid gland, a radioactive isotope of iodine, ^{131}I , is given by mouth in the form of sodium iodide.

(a) (i) State the position of the thyroid gland in the body.

..... [1]

(ii) Describe **one** method by which iodine ions may pass from the blood into the cells of the thyroid gland.

.....

 [3]

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

Within two days, a healthy thyroid gland takes up approximately 20% of a dose of ^{131}I given by mouth. The proportion taken up varies with the health and activity of the thyroid gland.

Fig. 4.1 shows the percentage uptake of ^{131}I by the thyroid gland in three people, **A**, **B** and **C**.

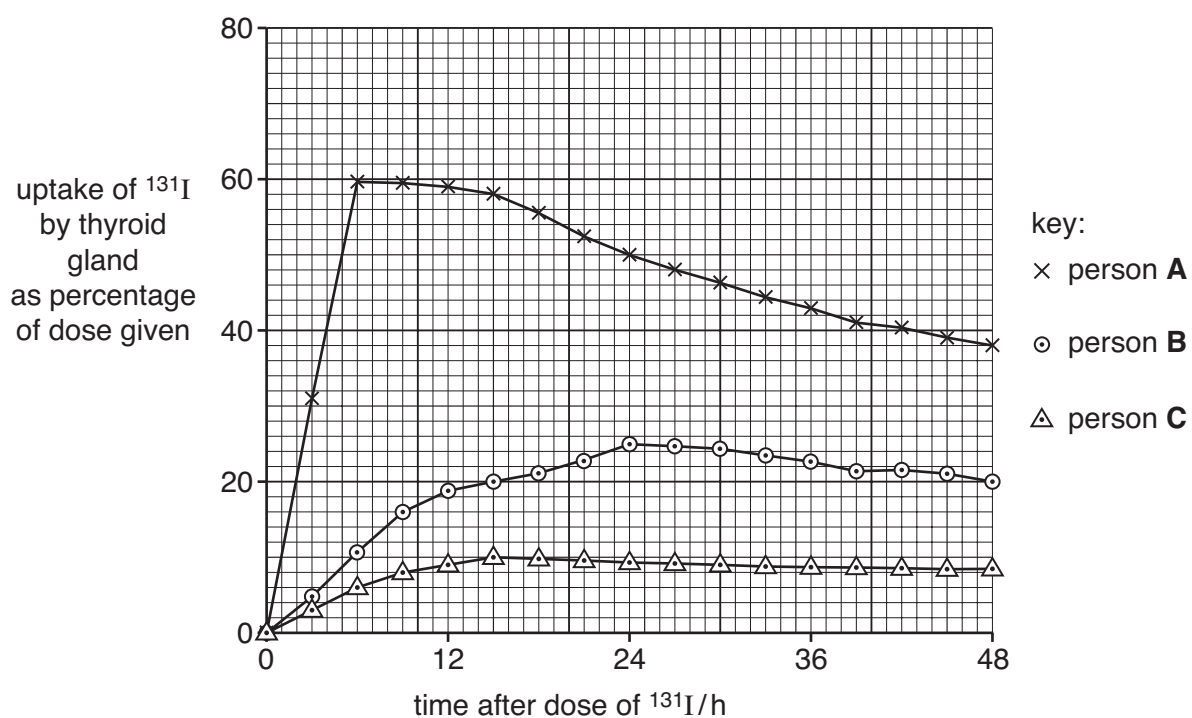


Fig. 4.1

[8]

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Turn over

- (c) If a person is exposed to a drop in the temperature of the environment for several days, the thyroid gland will increase its production of thyroxine. This results in an increase in body temperature.

- (i) Explain how thyroxine increases body temperature.

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..... [3]

- (ii) Suggest why thyroxine only achieves an increase in body temperature **over several days**.

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..... [1]

[Total: 17]

- (a) (i)** Osteoporosis and osteoarthritis are both degenerative diseases of the skeletal system.

Here are some statements about these diseases.

A	Bones may fracture.
B	Bone density decreases.
C	Cartilage breaks down.
D	Cigarette smoking increases the risk.
E	Synovial fluid decreases.
F	Caused by 'wear and tear'.
G	Cartilage becomes rough.
H	Hormonal cause.

Using the letters **A** to **H**, select the correct statements that apply to each of the degenerative diseases, osteoporosis and osteoarthritis.

The first one in each case has been done for you.

osteoporosis H

osteoarthritis C

[2]

- (ii) Suggest what lifestyle choices a young adult can make to **decrease** their chances of developing osteoporosis or osteoarthritis as they age.

..... [4]

(b) Rheumatoid arthritis is another disease that affects the skeletal system. This is an autoimmune disease in which the immune response destroys the lining of the person's **own** joints. The disease may be genetic in origin.

(i) Explain how the immune response could destroy the lining of a joint.

[5]

(ii) Suggest why movement of a joint is restricted following an autoimmune response.

..... [2]

(iii) Suggest how the rheumatoid arthritis form of autoimmunity may have developed.

..... [2]

(c) Discuss the **social** consequences of the effect of ageing on the skeletal system.

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..... [4]

[Total: 19]

- 6 Some people have a genetic form of total deafness from birth. Others may experience a progressive hearing loss as they grow older. However, many people retain good hearing into old age.

- (a) Investigations have shown that people with an increased susceptibility to hearing loss may possess a mutated form of a gene on chromosome 1, called KCNQ4.

KCNQ4 contains the genetic code for a potassium ion channel. These channels are found in the cell surface membranes of receptor cells in the ear, and in the cell surface membranes of neurones between the ear and the brain.

Fig. 6.1 shows part of the cell surface membrane in a neurone.

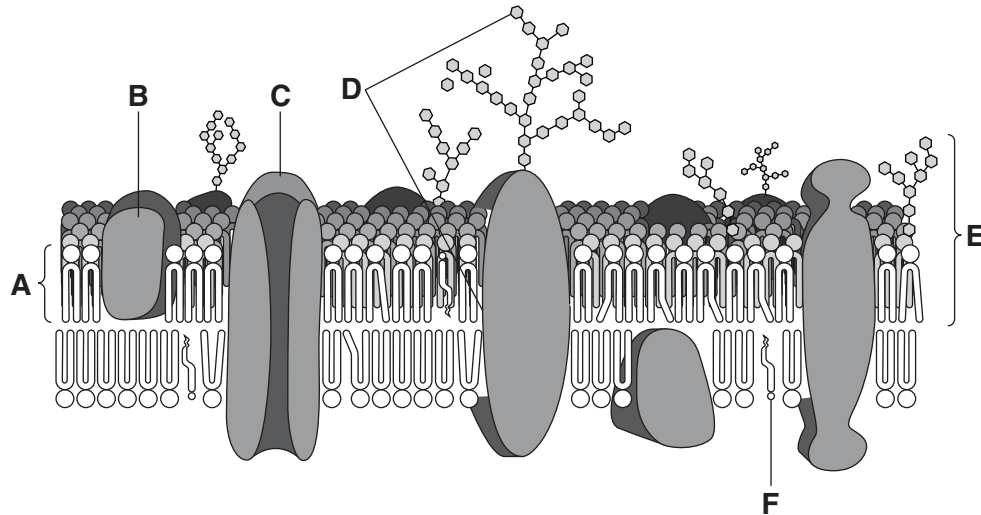


Fig. 6.1

- (i) State the letter on Fig. 6.1 that indicates a potassium ion channel.

..... [1]

- (ii) What is the potassium ion channel made of?

..... [1]

- (iii) Explain what is meant by the statement 'KCNQ4 contains the genetic code for a potassium ion channel'.

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..... [3]

- (iv) The mutation in gene KCNQ4 causes a change in the shape of the potassium ion channels.

Suggest how the mutation will affect the functioning of these channels in a neurone.

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..... [4]

Credit will be given for appropriate use of genetic diagrams.

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- (c) Single base mutations within a gene such as KCNQ4 can be observed by using the technique of DNA sequencing.

Fig. 6.2 shows the base sequence analysis of a normal part of the DNA making up a gene and the same part where the DNA has mutated.

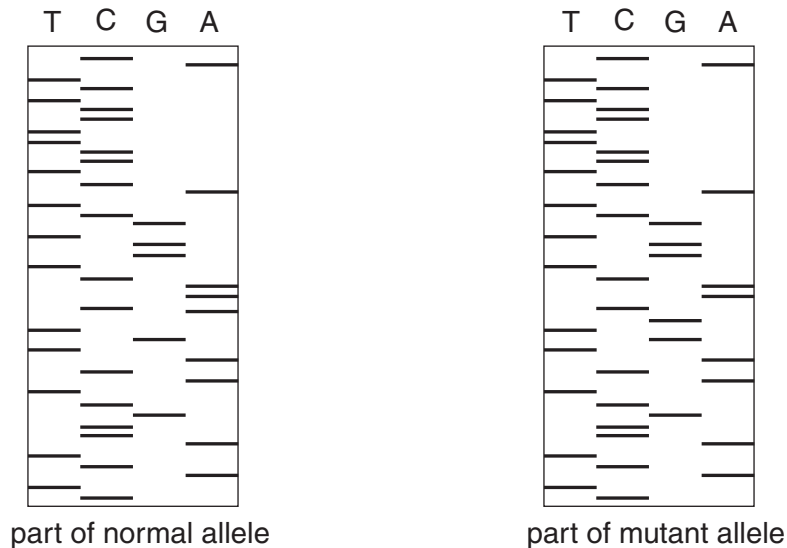


Fig. 6.2

From the sequences in Fig. 6.2, identify:

- (i) the base that is missing from the mutant allele;

..... [1]

- (ii) the base that has been substituted in its place.

..... [1]

- (iii) Many genetic abnormalities are caused by the deletion of a base. For example, cystic fibrosis may be caused by the deletion of a base in the CFTR gene on chromosome 7.

Explain why single base deletions usually have a more serious effect than base substitutions.

.....

 [3]

[Total: 22]

Turn over

- 7 Throughout their life, women will experience significant changes in their reproductive system that will affect their fertility.

(a) Fig. 7.1 shows the percentage occurrence of three different types of menstrual cycle depending on the age of the women.

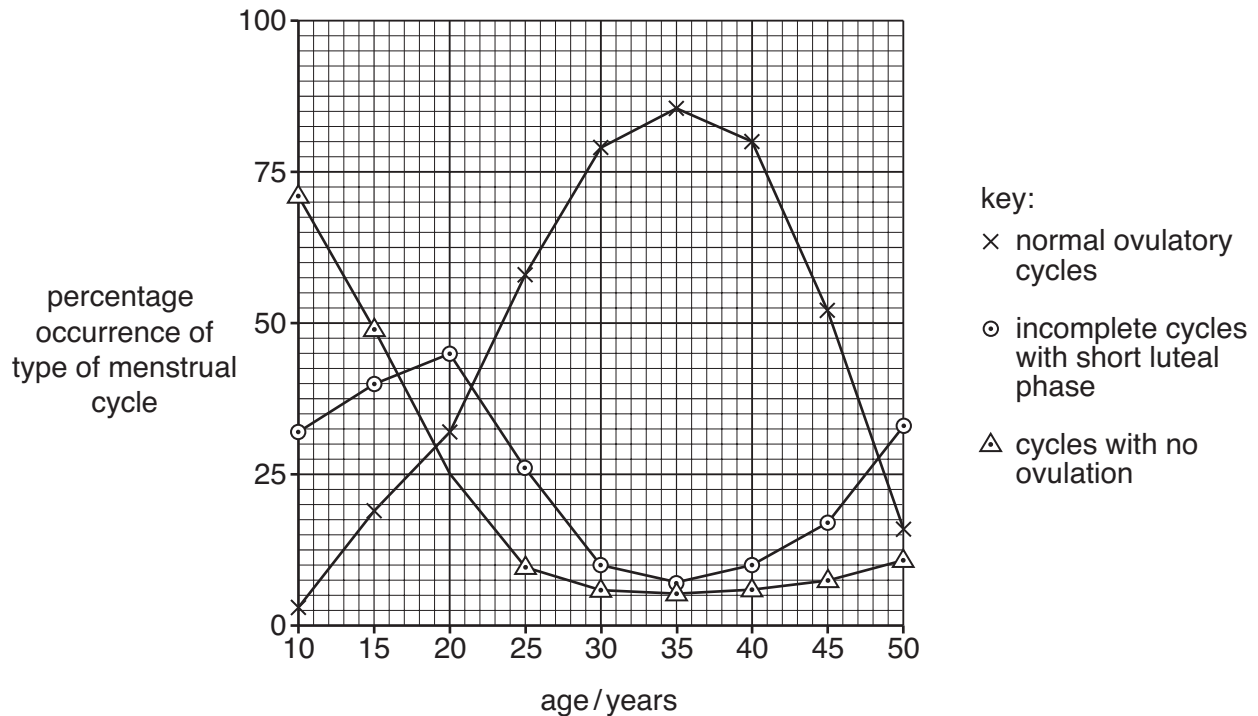


Fig. 7.1

- (i) Using the data in Fig. 7.1, describe the differences in the **type** of menstrual cycles in **35-year-old** women and **50-year-old** women.

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..... [4]

- (ii) Explain the change in **normal ovulatory cycles** between 35-year-old and 50-year-old women.

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..... [4]

- (iii) Explain the link between abnormal menstrual cycles and the development of osteoporosis in older women.

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..... [2]

- (iv) Suggest the likely effect of a short luteal phase.

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..... [3]

- (b) Suggest why the changes in the menstrual cycles of women between the ages of 35 and 50 years have developed during human evolution.

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..... [3]

- (c) It may be possible to **extend** the reproductive life of a woman by treating her with reproductive hormones.

Suggest **two** possible risks of treating an older woman with reproductive hormones.

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..... [2]

[Total: 18]

END OF QUESTION PAPER

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