

Tuesday 31 January 2012 – Afternoon

A2 GCE HUMAN BIOLOGY

F225 Genetics, Control and Ageing



Candidates answer on the Question Paper.

OCR supplied materials:

None

Duration: 2 hours

Other materials required:

- Electronic calculator
- Ruler (cm/mm)



Candidate forename						Candidate surname					
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Centre number							Candidate number				
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INSTRUCTIONS TO CANDIDATES

- Write your name, centre number and candidate number in the boxes above. Please write clearly and in capital letters.
- Use black ink. HB pencil may be used for graphs and diagrams only.
- Answer **all** the questions.
- Read each question carefully. Make sure you know what you have to do before starting your answer.
- Write your answer to each question in the space provided. If additional space is required, you should use the lined page at the end of this booklet. The question number(s) must be clearly shown.
- Do **not** write in the bar codes.

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 **100**.
-  Where you see this icon you will be awarded marks for the quality of written communication in your answer.
- You may use an electronic calculator.
- You are advised to show all the steps in any calculations.
- This document consists of **28** pages. Any blank pages are indicated.

Answer **all** the questions.

- 1 There are two types of motor neurones, somatic motor neurones and autonomic motor neurones.

Fig. 1.1(a) is a diagram of a somatic motor neurone. Fig. 1.1(b) shows an autonomic motor neurone.

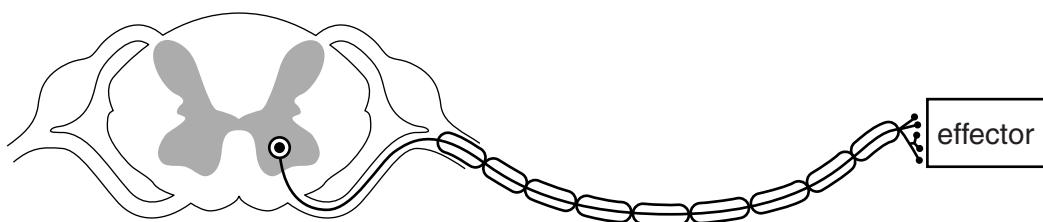


Fig. 1.1(a)

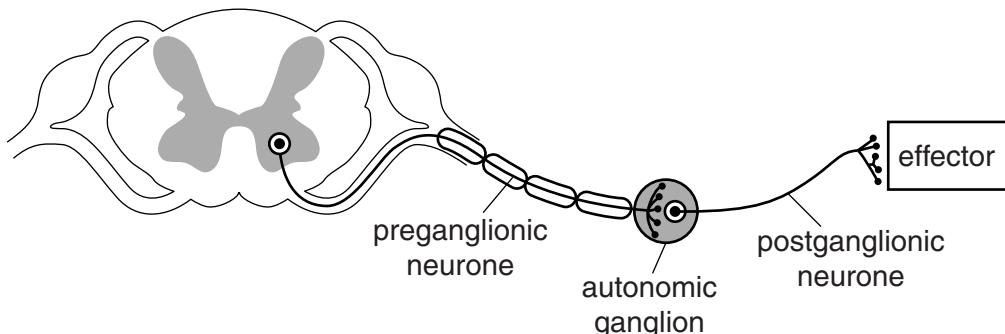


Fig. 1.1(b)

- (a) Somatic motor neurones have a myelinated axon.

- (i) Outline the function of the myelin sheath in the transmission of nerve impulses in **somatic** motor neurones.

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[2]

- (ii) With reference to Fig. 1.1(b), describe the distribution of the myelin sheath in **autonomic** motor neurones.

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[2]

- (iii) State **one** similarity and **one** difference between somatic and autonomic motor neurones, other than the distribution of myelin sheath.

similarity

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difference

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[2]

- (b) Autonomic motor neurones regulate the contraction of cardiac muscle in order to maintain supplies of oxygen and glucose to the tissues of the body. This is an important part of homeostasis.

Outline how the **autonomic nervous system** acts on cardiac muscle to regulate the supply of oxygen and glucose to the tissues.



In your answer you should refer to both branches of the autonomic nervous system.

[7]

[Total: 13]

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QUESTION 2 STARTS ON PAGE 6

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- 2 Fig. 2.1 is a diagram of a section through the human eye. Part of this section has been enlarged to show greater detail. The arrows indicate the flow of aqueous humour from where it is formed in the ciliary body to where it drains through the outflow channel.

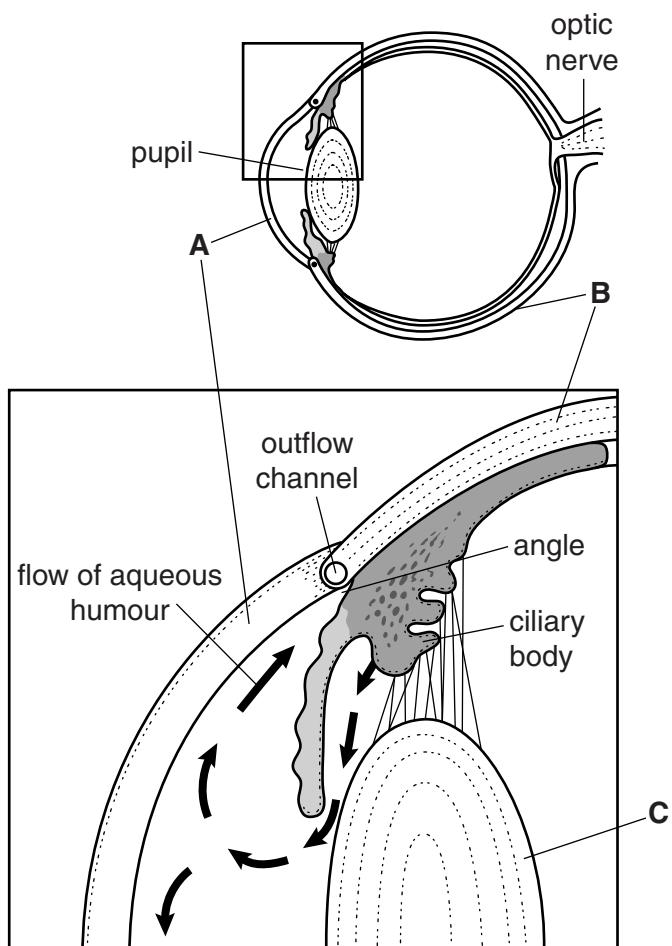


Fig. 2.1

- (a) Name the structures labelled **A**, **B** and **C**.

structure	name
A	
B	
C	

[3]

- (b) Glaucoma occurs when the aqueous humour fails to drain properly. This fluid builds up, raising the pressure in the eye. This may lead to blindness.

There are two forms of glaucoma:

- open angle glaucoma
- closed angle or acute glaucoma.

- (i) Suggest what is meant by the term 'acute' as used in the context of acute glaucoma.

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..... [1]

- (ii) Outline the effects on vision caused by glaucoma which, if left untreated, may cause blindness.

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..... [2]

- (c) Table 2.1 shows some of the causes of blindness in different regions of the world.

The figures are expressed as a percentage of the total number of blind people in each region.

Table 2.1

economic status	region	cause of blindness (percentage of total number of blind people per region)			
		cataract	glaucoma	retinal damage due to diabetes	age-related macular degeneration (AMD)
developed	United States of America	5	18	17	50
developing	South America	40	15	7	5
developed	Northern Europe	5	18	17	50
developing	Africa	55	15	0	0

- (i) Using the information in Table 2.1, describe how the causes of blindness differ between the developed and the developing regions of the world.

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[4]

- (ii) Suggest why there are marked differences in different regions of the world in the percentage of blindness due to cataracts.

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[2]

- (iii) Discuss whether the data in Table 2.1 supports the following statement:

"Type 2 diabetes is the main cause of retinal damage in diabetics".

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[2]

- (iv) Table 2.1 indicates that there are fewer recorded cases of age-related macular degeneration (AMD) in Africa than in other regions of the world. Suggest why.

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[2]

[Total: 16]

10

- 3 Chromosome mutations can result from non-disjunction during meiosis.

Fig. 3.1(a) and Fig. 3.1(b) show two different human karyotypes.

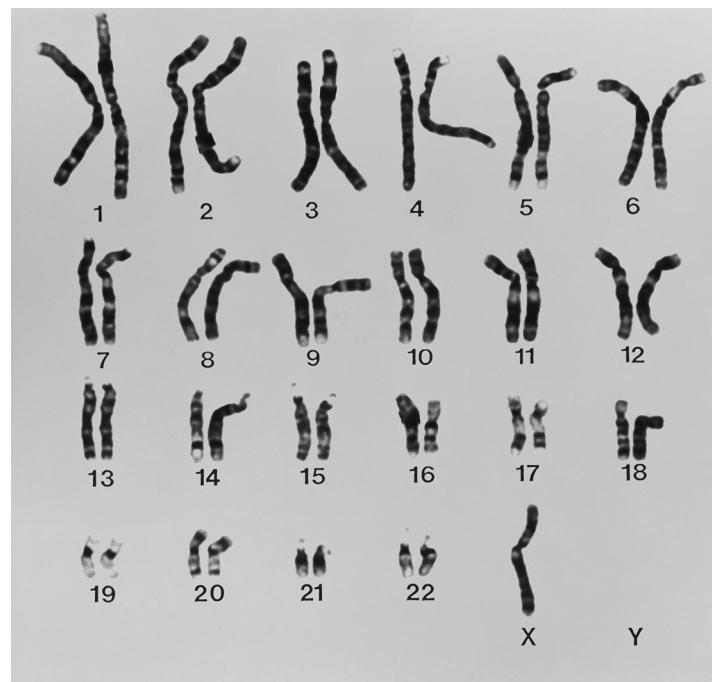


Fig. 3.1(a)

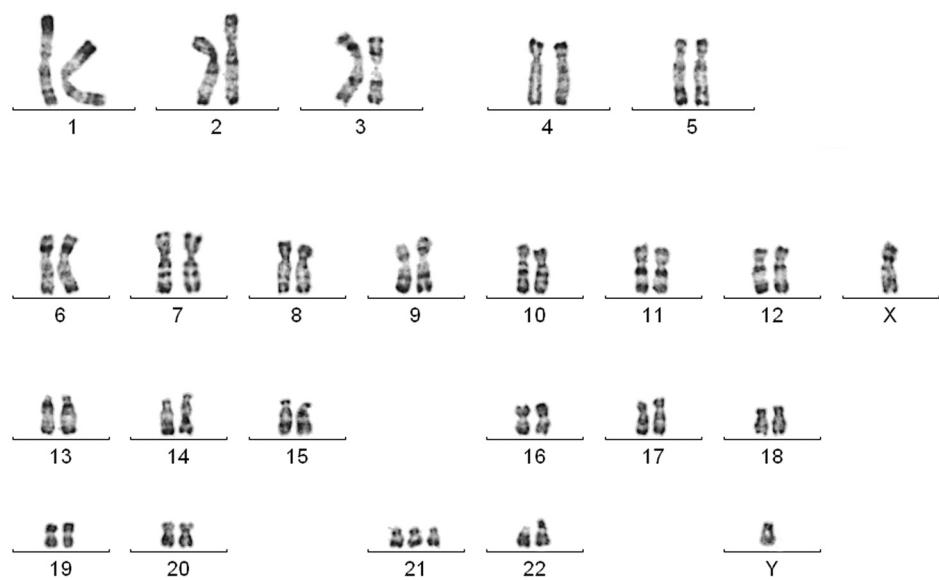


Fig. 3.1(b)

- (a) Fig. 3.1(a) is a karyotype from a girl with Turner's syndrome.

Identify, with reasons, the karyotype shown in Fig. 3.1(b).

[4]

[4]

- (b) Turner's syndrome is the most common chromosome mutation in females. Turner's syndrome can occur due to non-disjunction in **meiosis** during gametogenesis. The resulting gamete will be missing an X chromosome.

The following statements all relate to different stages of gametogenesis.

	statement
D	a primary spermatocyte divides to form secondary spermatocytes
E	a germinal epithelial cell divides to produce oogonia
F	a primary oocyte divides to produce a secondary oocyte
G	a secondary spermatocyte divides to produce spermatids
H	a spermatogonium divides to produce primary spermatocytes
J	a germinal epithelial cell divides to produce spermatogonia

Using the letters, **D** to **J**, select **three** statements which identify a stage where non-disjunction in meiosis could result in a gamete with a missing X chromosome.

statement letters

.....

[3]

- (c) Girls that are diagnosed with Turner's syndrome are often placed under the care of an endocrinologist who supervises a programme of hormone treatment.

 - Treatment with **human growth hormone** begins between 2 and 5 years of age.
 - Treatment with **oestrogen** replacement therapy begins between 12 and 15 years of age.

Discuss why the hormone treatments described above could be considered to be both ethical and unethical.



In your answer you should give a balanced discussion of both the ethical and the unethical aspects of these treatments.

[6]

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QUESTION 3(d) STARTS ON PAGE 14

- (d) Some forms of Turner's syndrome occur when one of the pair of X chromosomes becomes damaged. The damaged X chromosome may have been broken and re-formed so that part of its structure is missing.

Fig. 3.2 is a diagram of a normal X chromosome and two forms of 'damaged' X chromosomes, Xp and Xq.

- In deletion Xp, a section of the 'p' arm of the chromosome is missing. This deletion leads to reduced height and abnormalities such as narrowing of the aorta.
- In deletion Xq, a section of the 'q' arm of the chromosome is missing. This deletion leads to little or no development of the gonads.

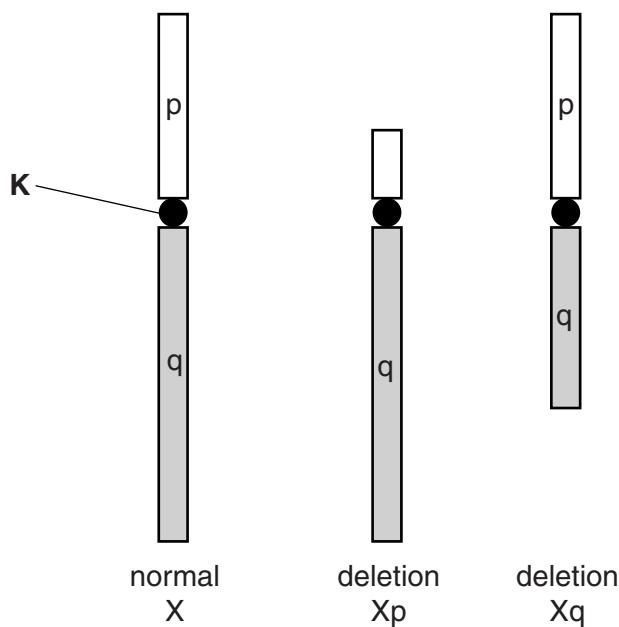


Fig. 3.2

- (i) Name structure K.
- [1]

- (ii) Explain why deletion Xp and deletion Xq result in different phenotypes.

.....

 [2]

- (e) Mothers with the Xp or Xq form of Turner's syndrome can pass on the chromosome mutation to their daughters but not to their sons.

- (i) Complete the genetic diagram below to show how the chromosome mutation Xp may be passed on to **daughters** from a mother with Turner's syndrome.

parental genotypes : XXp

parental phenotype:	female with Turner's syndrome	normal male
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possible gametes

possible genotypes
of daughters

possible phenotypes
of daughters

[4]

- (ii) Suggest why the Xp form of Turner's syndrome is not inherited by male offspring.

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 [2]

[Total: 22]

- 4 Alpha-1-antitrypsin (AAT) is a glycoprotein that is produced naturally by liver cells. AAT acts as an inhibitor of protease enzymes. One example of a protease is the enzyme **elastase**, which is produced by phagocytic cells in lung tissue in response to damage. The role of AAT is to protect the healthy tissues from attack by these enzymes.

The gene for AAT has been genetically engineered and inserted into sheep cells so that the glycoprotein is produced in the cells of the mammary glands and secreted into the sheep's milk.

- (a) Table 4.1 is a summary of the steps involved in genetic engineering using sheep cells for the production of AAT. The steps are not in the correct order.

Table 4.1

letter	step in procedure
A	Sheep cells treated with vector and grown on antibiotic to detect cells that successfully take up vector (transformed cells).
B	Vector prepared containing human gene coding for production of AAT, a promoter sequence and a gene for antibiotic resistance.
C	Offspring treated with hormones and the milk produced tested for presence of AAT.
D	Embryo implanted in uterus of sheep.
E	Transformed cells fused with sheep egg cell from which nucleus has been removed and cells start to divide.
F	Sheep cells are isolated and grown in tissue culture medium.

- (i) Using the letters, place the steps in the correct order.

The first letter has been entered for you.

F

[3]

- (ii) Name the enzyme needed to attach different parts of the vector to each other as described in statement **B**.

..... [1]

- (iii) The promoter sequence in statement **B** is a region of DNA to which the enzyme RNA polymerase can bind so that mRNA can be synthesised.

Suggest why the promoter sequence chosen for AAT production was a promoter for the synthesis of a milk protein.

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[1]

- (iv) Suggest which **two** hormones could be used in statement **C**.

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[2]

- (b) Alpha-1-antitrypsin deficiency (AATD) is a condition caused by a recessive mutant allele. Children who inherit two copies of this allele are unable to produce AAT.

Parents who know they are carriers of the mutant allele may seek advice from a genetic counsellor.

Discuss briefly the **ethical** reasons why genetic engineering of human cells using the technique described in Table 4.1 would **not** be an option that the genetic counsellor could advise.

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[3]

- (c) Two symptoms of AATD are pulmonary emphysema and liver cirrhosis.

Treatment of pulmonary emphysema is possible in some patients using AAT injections.

For patients in the later stages of the disease, the only effective treatment is organ transplant surgery.

- (i) Suggest one further cause of pulmonary emphysema and one further cause of liver cirrhosis in people **without AATD**.

pulmonary emphysema

liver cirrhosis [2]

- (ii) Describe how genetic engineering has the potential to provide **non-human** organs for transplantation. Discuss **briefly** why some people may object to the use of such organs.

[7]

[Total: 19]

QUESTION 5 STARTS ON PAGE 20

- 5 Cannabis is produced by the plant *Cannabis sativa*. Cannabis contains a compound called THC which has been shown to affect brain function. THC can act as a pain killer and a sedative. For this reason, cannabis has been prescribed by some doctors for therapeutic purposes.

- (a) (i) Suggest what is meant by the term 'therapeutic'.

.....
.....

[1]

- (ii) Suggest **two** conditions for which cannabis could be used therapeutically.

1.

2. [2]

- (b) Despite being an illegal drug in the UK, cannabis is one of the most commonly used recreational drugs.

- It is possible to detect the use of cannabis since breakdown products of THC, such as **THC carboxylic acid**, are present in blood plasma and are excreted in urine.
- One dose of cannabis can be detected for up to 6 days in some people.
- Frequent users can still test positive for THC carboxylic acid for 30 days or more after last using cannabis.

A study was carried out to investigate the presence of breakdown products of THC in the urine of two volunteers. Both volunteers smoked one cigarette containing the same quantity of cannabis. Their urine was tested at intervals for the next 150 hours.

The results are shown in Fig. 5.1.

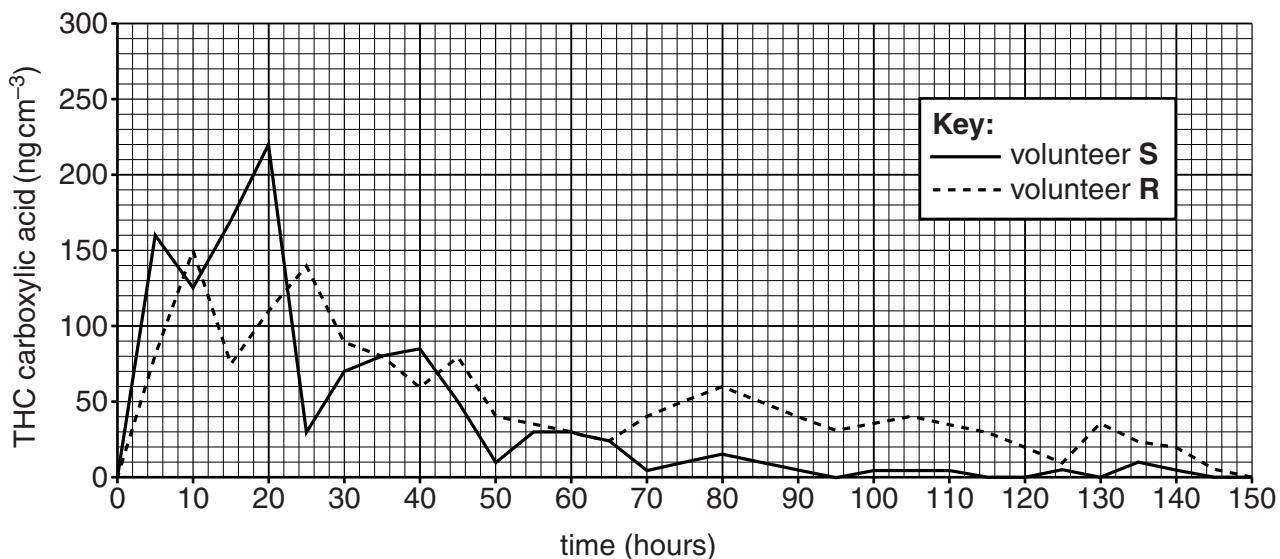


Fig. 5.1

- (i) THC carboxylic acid can be detected in urine using test kits which vary in their accuracy of detection. The lowest concentration at which THC carboxylic acid could be detected in the study in Fig. 5.1 was 50 ng cm^{-3} .

Using the information in Fig. 5.1, state the time **beyond which** volunteers R and S would **not** have tested positive for cannabis using this test kit.

R _____

S

[2]

- (ii) Outline how breakdown products from drugs, such as THC, pass from blood plasma into urine.

[4]

- (iii) Suggest why the concentration of the THC breakdown product rises and falls in urine samples, as shown in Fig. 5.1.

[2]

[2]

QUESTION 5(c)(i) STARTS ON PAGE 22

- (c) (i) Suggest reasons why the measurement of THC carboxylic acid in urine is **not** a good indicator of cannabis **intoxication**.

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.....

[2]

- (ii) State **two** symptoms of cannabis intoxication.

.....
.....

[Total: 15]

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QUESTION 6 STARTS ON PAGE 24

- 6 Following the prolonged period of extreme cold weather in the winter of 2009–2010 in the UK, several experts predicted a rise in the number of deaths due to hypothermia.

- (a) State precisely what is meant by the term hypothermia.

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.....
.....

[2]

- (b) Complete the following table to distinguish between moderate and severe hypothermia.

The first row has been completed for you.

symptom	moderate hypothermia	severe hypothermia
mental state	confusion	unable to communicate
shivering		
breathing rate		

[2]

- (c) The impact of winter on mortality is measured by calculating a figure for excess winter mortality (EWM).

This figure is calculated by comparing deaths occurring from December to March with the average number of deaths at other times of the year.

In the winter period of 2008–2009, the EWM figure in the UK was 36700. This was an increase of 49% compared to the EWM for the previous year (2007–2008).

- (i) Calculate the EWM for the year 2007–2008.

Show your working.

Give your answer to the nearest whole number.

Answer = deaths [2]

- (ii) A lowering of body temperature can result in an increase in blood viscosity and a rise in blood pressure.

Suggest why these clinical signs mean that **elderly** people are **more** at risk of dying from hypothermia.

[Total: 10]

- 7 One symptom of the onset of menopause is rapid and sudden changes in body temperature known as 'hot flushes'.

Hot flushes can be embarrassing and uncomfortable and are one reason why some women choose to have hormone replacement therapy (HRT).

There are three main types of HRT:

- continuous combined HRT
- combined HRT
- cyclic HRT.

Fig. 7.1 is a diagram showing how hormones are given during two monthly cycles for the three types of HRT.

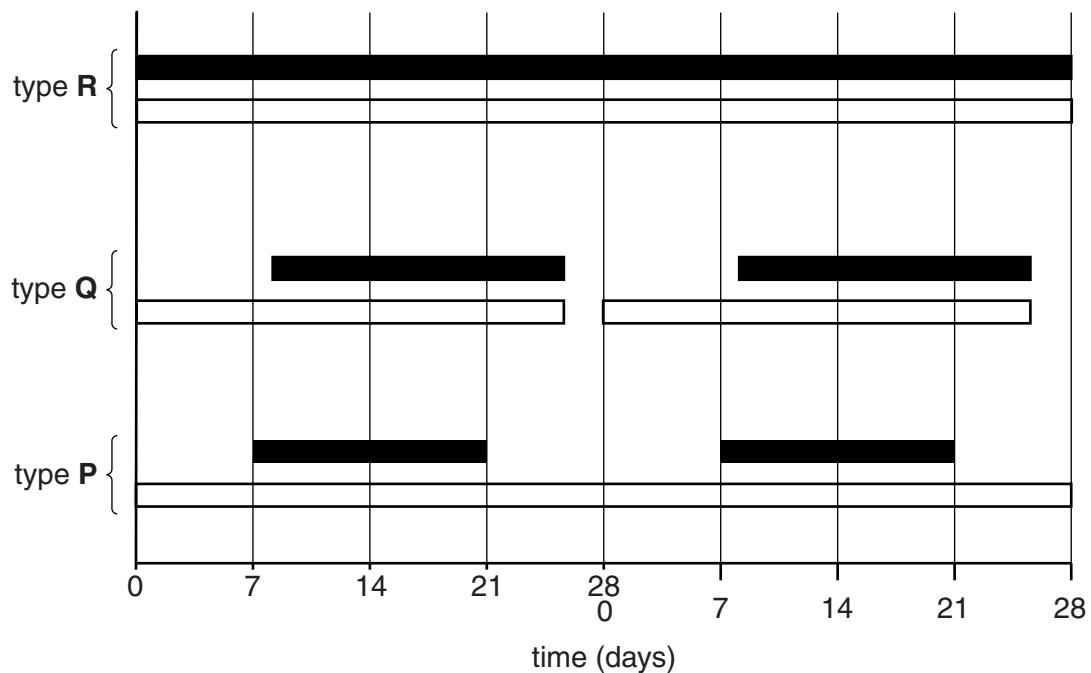


Fig. 7.1

- (a) Identify the **two** hormones by writing the name of each hormone alongside the key to Fig. 7.1 shown below.

Key to hormones in Fig. 7.1:

[Redacted]

[Redacted]

[2]

- (b) Identify which type of HRT is shown by the letters **P, Q and R** in Fig. 7.1.

type of HRT	letter
continuous combined HRT	
combined HRT	
cyclic HRT	

[3]

[Total: 5]

END OF QUESTION PAPER

ADDITIONAL PAGE

If additional space is required, you should use the lined page below. The question number(s) must be clearly shown.



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