

**OXFORD CAMBRIDGE AND RSA EXAMINATIONS**  
**A2 GCE**  
**F225/01**

**HUMAN BIOLOGY**  
**Genetics, Control and Ageing**

**WEDNESDAY 17 JUNE 2015: Morning**

**DURATION: 2 hours**  
**plus your additional time allowance**

**MODIFIED ENLARGED**

<b>Candidate forename</b>		<b>Candidate surname</b>	
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<b>Centre number</b>						<b>Candidate number</b>				
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**Candidates answer on the Question Paper.**

**OCR SUPPLIED MATERIALS:**

**None**

**OTHER MATERIALS REQUIRED:**

**Electronic calculator**

**Ruler (cm/mm)**

**READ INSTRUCTIONS OVERLEAF**

## **INSTRUCTIONS TO CANDIDATES**

**Write your name, centre number and candidate number in the boxes on the first page. 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 pages at the end of this booklet. The question number(s) must be clearly shown.**

## **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.**

**Any blank pages are indicated.**

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**Answer ALL the questions.**

**1 Homeostasis has been defined as ‘the maintenance of a *constant* or *stable* internal environment’.**

**(a) Explain why the word ‘stable’ is more appropriate than the word ‘constant’ in a definition of homeostasis.**

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**[2]**

**(b) The autonomic nervous system plays an important role in homeostasis.**

**The terms listed below could be used to describe the features or activity of the two branches of the autonomic nervous system.**

<b>ACETYLCHOLINE</b>	<b>ADRENALINE</b>	<b>DECREASE</b>
<b>GLYCOGENESIS</b>	<b>GLYCOGENOLYSIS</b>	<b>INCREASE</b>
<b>NORADRENALINE</b>	<b>MOTOR</b>	<b>SENSORY</b>

**Complete Table 1.1 below by inserting the most appropriate term.**

**Terms may be used once, more than once or not at all.**

**Some boxes have been completed for you.**

**TABLE 1.1**

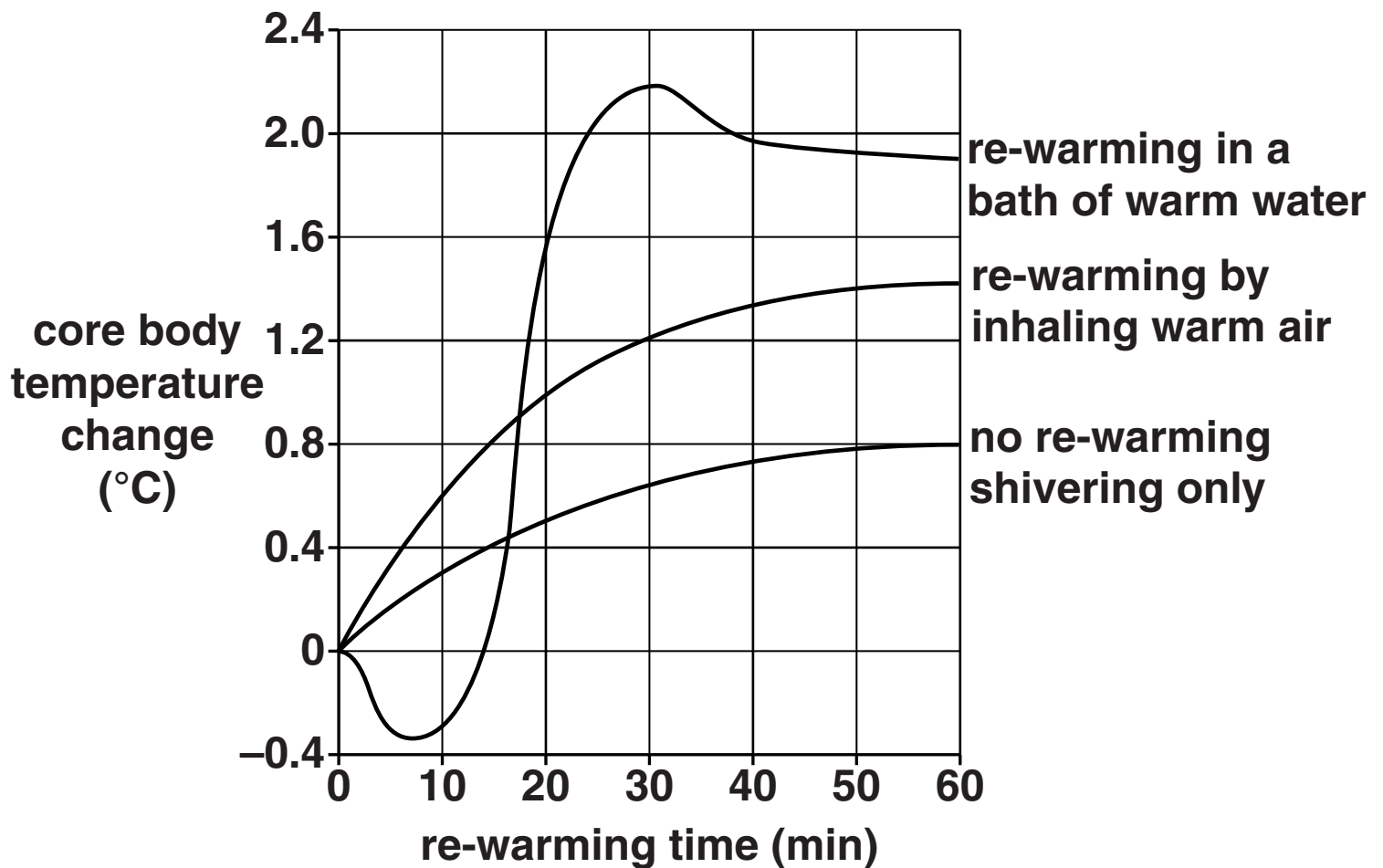
<b>FEATURE OR ACTIVITY</b>	<b>BRANCH OF AUTONOMIC NERVOUS SYSTEM</b>	
	<b>PARASYMPATHETIC</b>	<b>SYMPATHETIC</b>
<b>Type of neurone</b>	<b>motor</b>	
<b>Process triggered in liver cells</b>		
<b>Effect on heart rate</b>		
<b>Neurotransmitter released at the SAN</b>		<b>noradrenaline</b>

**[4]**

- (c) Control of body temperature is an important aspect of homeostasis. Conditions such as hypothermia can be fatal if not treated.

Fig. 1.1 compares the effects of three methods of treatment for mild hypothermia.

FIG. 1.1



- (i) Explain how shivering causes a rise in core body temperature.**

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**[3]**

- (ii) Fig. 1.1 opposite shows that placing a person with hypothermia in a bath of warm water results in an initial drop in CORE body temperature.**

**Suggest WHY the core body temperature drops initially.**

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**[2]**

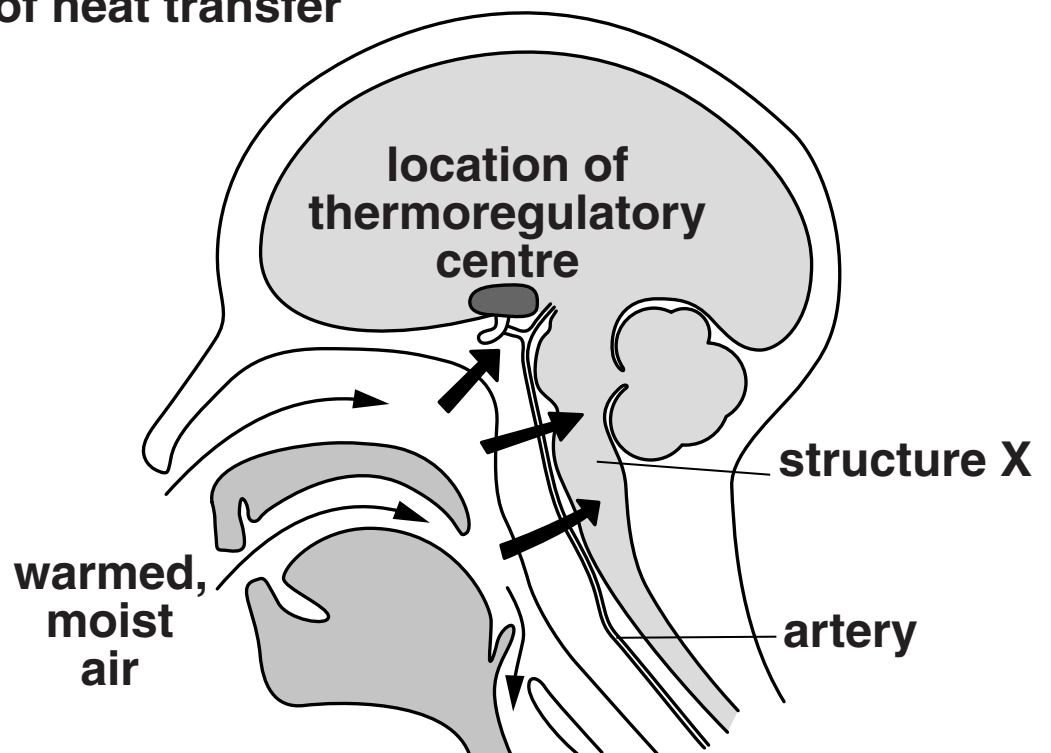
- (d) Re-warming by inhaling warmed air is also known as **CORE RE-WARMING**. Fig. 1.2 shows the route taken by the warm air as it is inhaled by the person.

**FIG. 1.2**

**KEY:**

→ direction of warm air movement

➔ direction of heat transfer



- (i) Name the part of the brain in which the thermoregulatory centre is located.

\_\_\_\_\_ [1]

- (ii) Identify structure X.

\_\_\_\_\_ [1]



- (iii) Using the information in Fig. 1.2 opposite, suggest why core re-warming is better than other methods of re-warming at treating severe hypothermia.

In your answer, you should refer to the symptoms of **SEVERE** hypothermia.

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[3]

[TOTAL: 16]

- 2 Down's syndrome (DS) is a genetic condition that occurs in approximately 1 in every 800 births.

FIG. 2.1a shows a karyotype from a male with a rare form of DS. In this karyotype, there is an additional piece of genetic material attached to one copy of chromosome 14.

A larger diagram of both copies of chromosome 14 is shown in FIG. 2.1b.

FIG. 2.1a

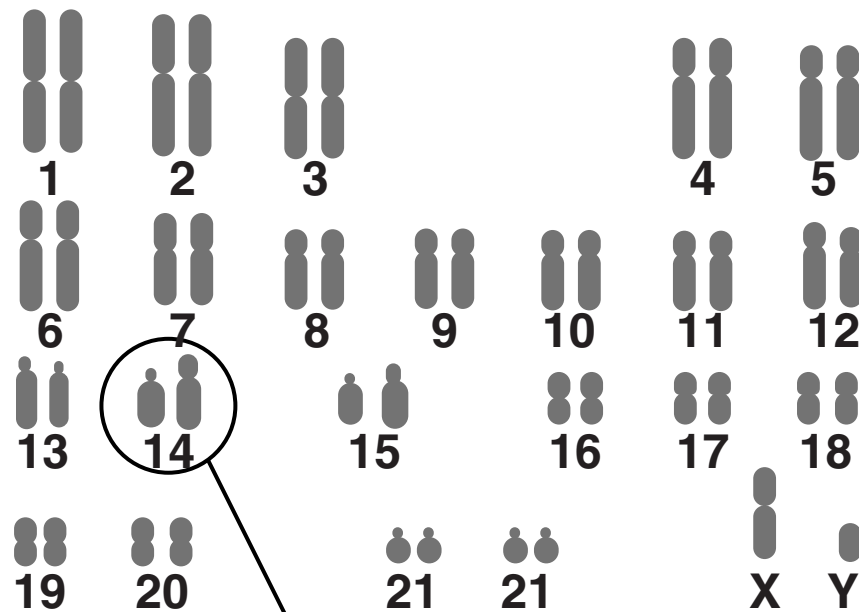
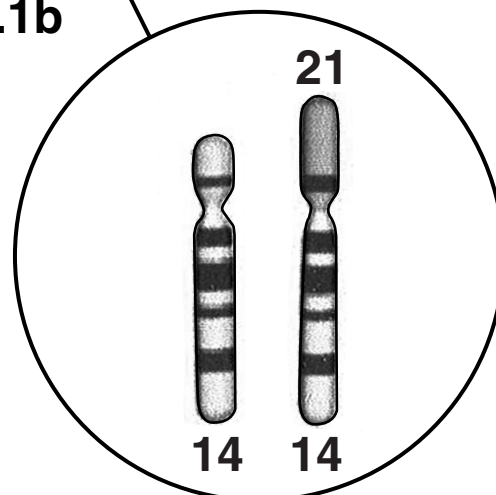


FIG. 2.1b



- (a) Describe how the appearance of the karyotype shown in Fig. 2.1a opposite differs from that of the more common form of DS.

Suggest WHEN and HOW the transfer of extra genetic material to chromosome 14 occurred, and what events led to cells with this karyotype being formed.



In your answer, you should include a comparison of the karyotypes and describe the events during and after meiosis that led to the formation of the karyotype in FIG. 2.1a.

[7]

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**(b) The National Cytogenetic Register for DS was set up in the United Kingdom in 1989.**

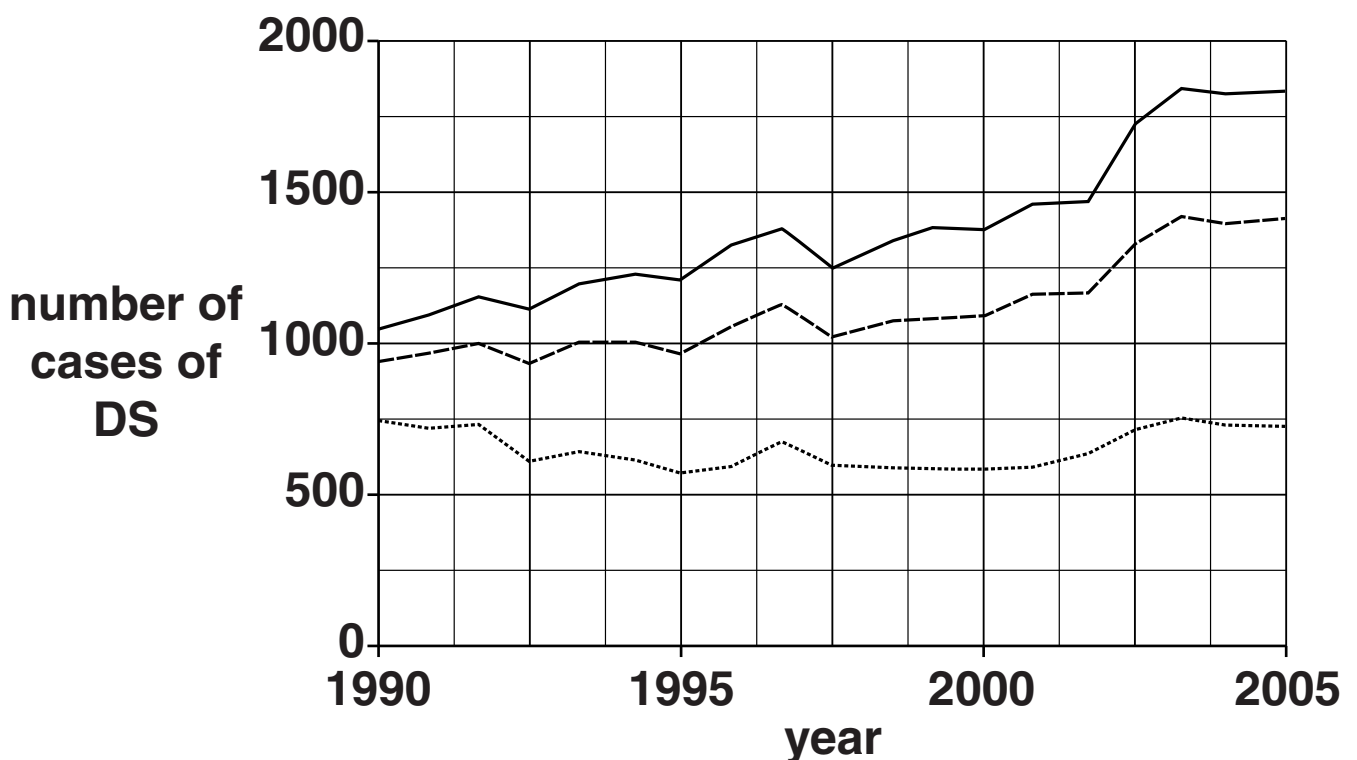
**This register holds anonymous data on families for over 26 000 cases of DS. These cases were diagnosed either antenatally or postnatally.**

**Fig. 2.2 shows the changes in the number of DS cases diagnosed between 1990 and 2005. It also shows the changes in the number of live births of children with DS for the same period.**

**FIG. 2.2**

**KEY:**

- total number of cases of DS diagnosed**
- estimated number of live births assuming no antenatal screening or terminations were available**
- ..... actual number of live births when antenatal screening or terminations were available**



- (i) Suggest why the data on the National Cytogenic Register for DS are held anonymously.**

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[1]

- (ii) Using the information in Fig. 2.2 opposite, suggest WHY the number of cases of DS DIAGNOSED between 1990 and 2005 has changed.**

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[2]

- (iii) Suggest what factor the investigators took into account in estimating the number of children with DS who would have been BORN, assuming that no antenatal screening or terminations were available.**

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[1]

**[TOTAL: 11]**

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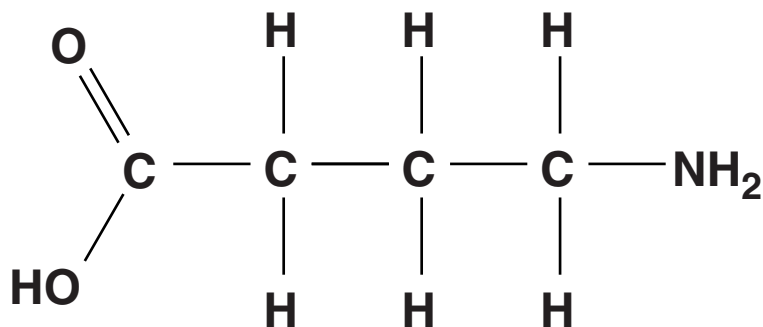


- 3 GABA (gamma-aminobutyric acid) is one of the most common neurotransmitters in the human central nervous system.**

**GABA is synthesised from the amino acid glutamate.**

**(a) Fig. 3.1 is a diagram of GABA.**

**FIG. 3.1**



**GABA could also be described as an amino acid.**

**ON FIG. 3.1, circle and name the TWO chemical groups on the GABA molecule which indicate that it is also an amino acid.**

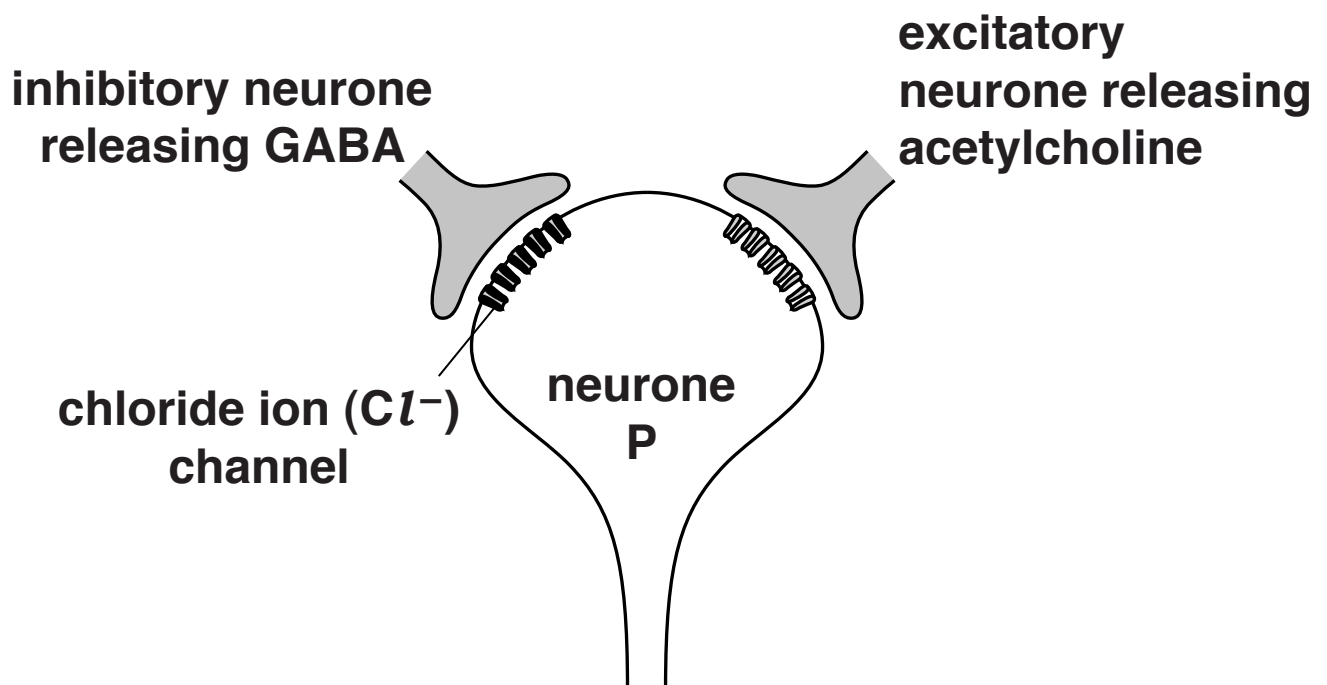
**The answer must be drawn on Fig. 3.1. [1]**

**(b) GABA acts as an INHIBITORY NEUROTRANSMITTER.**

**When GABA binds to its receptor, a chloride ion channel is opened and chloride ions ( $\text{Cl}^-$ ) enter the neurone.**

**Fig. 3.2 represents two neurones forming synapses with a third neurone.**

**FIG. 3.2**



**Suggest how GABA is released from the inhibitory neurone AND, using the information in Fig. 3.2 opposite, explain why an action potential may not occur in neurone P if both GABA and acetylcholine are released.**



**In your answer, you should make clear both the sequence of events in the release of the neurotransmitter and why an action potential could be prevented by GABA.** [6]

[illegible]

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- (c) GABA is the neurotransmitter in the parts of the brain responsible for sensations such as fear and anxiety. These sensations result from neurones becoming OVER-STIMULATED.**

**Diazepam is a drug which is often prescribed to reduce anxiety. This drug can also be used to reduce alcohol withdrawal symptoms and muscle spasms.**

- (i) State TWO symptoms of alcohol withdrawal OTHER THAN anxiety.**

**1 \_\_\_\_\_**  
**2 \_\_\_\_\_**  
**[1]**

- (ii) Name ONE OTHER condition for which Diazepam could be used therapeutically.**

**\_\_\_\_\_ [1]**

**[TOTAL: 9]**

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**4 Some aspects of human physiology, behaviour and mental activity follow a pattern which cycles over a period of approximately 24 hours. This cyclical pattern is called the ‘circadian rhythm’. It coincides approximately with periods of light and darkness.**

**(a) Circadian disorders, such as sleep-wake cycle disturbances, are associated with normal ageing.**

**What problems might elderly people or their carers experience as a result of sleep-wake disturbances?**

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**[3]**

**(b) Circadian rhythm is regulated as follows:**

**a part of the brain called the SCN receives impulses from the OPTIC NERVE**

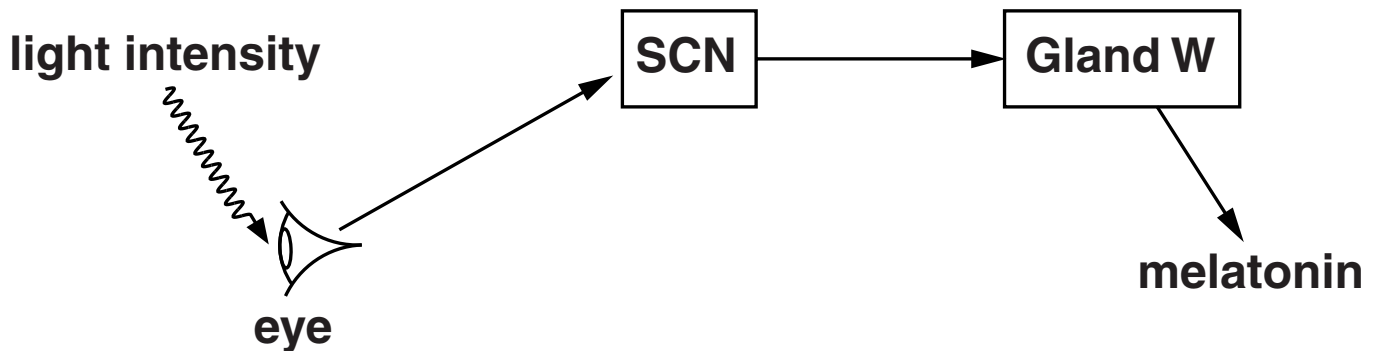
**the SCN regulates a gland in the brain which secretes a substance called MELATONIN**

**melatonin levels in blood are lowest during the day and highest at night**

**high levels of melatonin induce inactivity and sleep.**

**Fig. 4.1 is a summary of the interaction between the SCN and melatonin.**

**FIG. 4.1**





**(i) What type of gland is W? \_\_\_\_\_**

**What type of substance is melatonin?**

**Give a reason for your answers.**

**[2]**

**(ii) Using the information in Fig. 4.1 opposite, suggest why circadian rhythm problems are more likely to occur in the following groups of people:**

**those who are housebound \_\_\_\_\_**

**those with untreated glaucoma \_\_\_\_\_**

**those with Alzheimer's. \_\_\_\_\_**

**[3]**

- (c) Studies of melatonin levels in blood show large variations in concentrations between individuals.**

**The size of gland W has also been shown to vary between individuals.**

- (i) Suggest a technique that could be used to allow the size of gland W to be measured SAFELY and REPEATEDLY.**

**Explain the reason for your suggestion.**

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**[2]**

- (ii) LONGITUDINAL STUDIES are studies of populations over time.**

**Researchers have suggested that a longitudinal study would be a better experimental design for investigating the link between ageing, melatonin and changes in circadian rhythms.**

**In such a study, identify the following:**

**the independent variable** \_\_\_\_\_

**a variable that WOULD BE controlled in a longitudinal study.**

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**[2]**

- (iii) A study showed a **NEGATIVE CORRELATION** between melatonin concentration in blood and ageing.

Complete the graph below to show this correlation.



[2]

[TOTAL: 14]

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- 5 The control of blood glucose is essential because changes in blood glucose concentration will change the water potential of blood.**

**Water potential changes are detected by the osmoregulatory centre and adjustments are then made to the secretion of ADH.**

- (a) (i) State ONE reason, other than the effect that it has on water potential, why blood glucose concentrations must be controlled.**

\_\_\_\_\_

\_\_\_\_\_

\_\_\_\_\_ **[1]**

- (ii) State the location in the brain of the osmoregulatory centre AND the source of ADH secretion.**

**osmoregulatory centre** \_\_\_\_\_

**source of ADH secretion** \_\_\_\_\_ **[1]**

**(b) If people with type 1 diabetes do not administer the appropriate medication, the concentration of glucose in the blood can remain higher than the normal range for relatively long periods of time.**

**(i) Name the medication that is required to treat type 1 diabetes.**

**Describe how and when this medication is administered AND how it acts to reduce the blood glucose concentration.**

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**[5]**

- (ii) **PREDICT** the effect of high blood glucose concentration on ADH release **AND** urine volume. You should assume that no glucose is lost from the blood and that glucose levels remain high. Justify your prediction.

effect on ADH release \_\_\_\_\_

effect on urine volume \_\_\_\_\_

justification \_\_\_\_\_

\_\_\_\_\_

\_\_\_\_\_

\_\_\_\_\_

\_\_\_\_\_

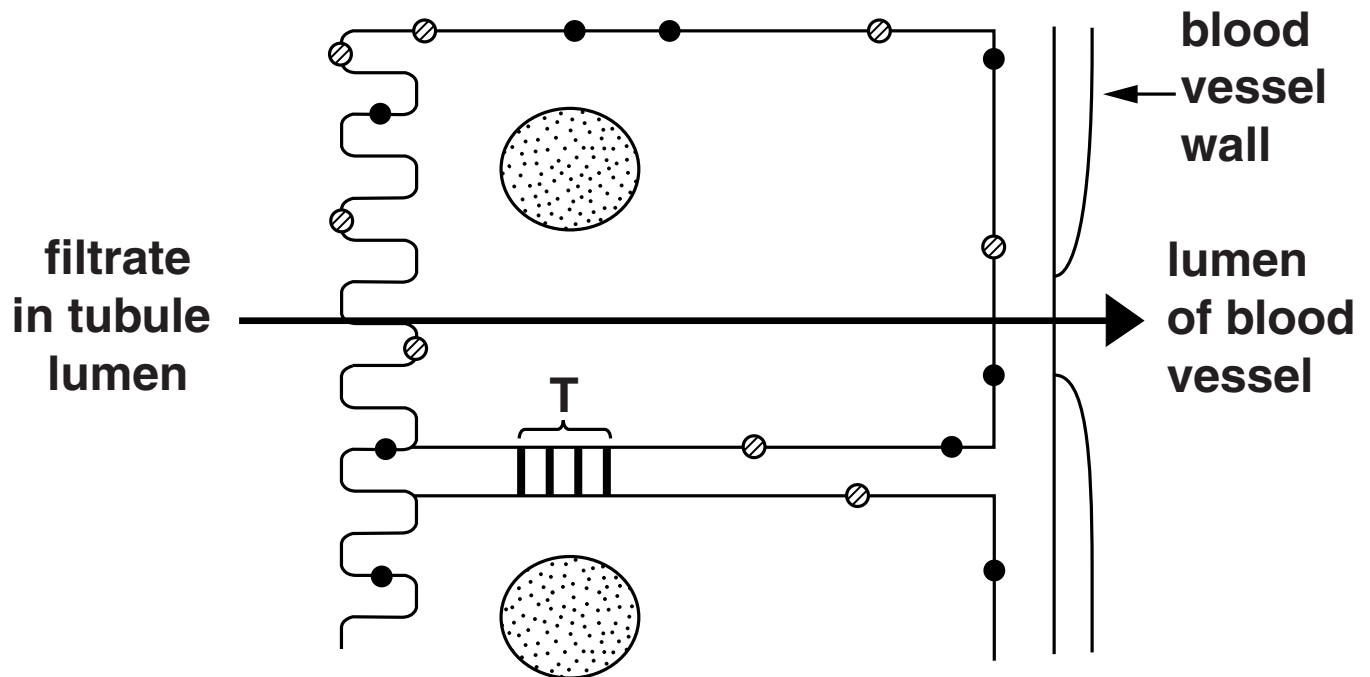
**[3]**

(c) Fig. 5.1 represents the structure of part of a kidney nephron and its surrounding blood vessels.

**FIG. 5.1**

**KEY**

- ⊙ carrier proteins
- channel proteins



(i) Using Fig. 5.1, identify, with a reason, the following:

the region of the nephron shown \_\_\_\_\_

reason \_\_\_\_\_

the type of blood vessel shown \_\_\_\_\_

reason \_\_\_\_\_

[2]



- (ii) Structure T is a ‘tight junction’ between two adjacent cells. Tight junctions prevent proteins located at the surface of cells from changing their positions.**

**Suggest why, if there were no tight junctions, proteins in cell surface membranes would be able to change positions.**

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**[2]**

- (iii) The region of the nephron shown in Fig. 5.1 opposite is the region where glucose is reabsorbed.**

**Using the information in Fig. 5.1, name a mechanism used for selective reabsorption of glucose AND explain why glucose appears in urine when blood glucose concentrations are high.**

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**[3]**

**(d) Some people with diabetes will develop a condition called diabetic kidney disease. One of the symptoms of this condition is the appearance of the protein albumin in the urine.**

**(i) Suggest where albumin is normally found in the body.**

\_\_\_\_\_ **[1]**

**(ii) Suggest, with reasons, which region of the kidney is damaged due to diabetic kidney disease.**

\_\_\_\_\_  
\_\_\_\_\_  
\_\_\_\_\_  
\_\_\_\_\_  
\_\_\_\_\_  
\_\_\_\_\_ **[2]**

**[TOTAL: 20]**

**6 The age at which a woman goes through the menopause depends on several factors.**

**Smoking, unemployment and being at higher risk of coronary heart disease are factors linked to a lower age of menopause.**

**Having children, use of oral contraception and having a lower BMI are factors linked to a higher age of menopause.**

**The age of menopause varies between different ethnic groups.**

**In a study involving large populations of women, the median age of 'natural menopause' in a population was found to be 50.4 years.**

**(a) (i) Suggest what is meant by natural menopause.**

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**[2]**

**(ii) A MEDIAN value for a large population is obtained as follows:**

**all the values obtained are placed in order from lowest to highest**

**the median is the middle value in the range.**

**Suggest why the MEAN age of menopause can vary significantly between populations but the MEDIAN does not.**

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**[2]**

- (b) Chemicals in cigarette smoke have been shown to cause damage to cells in ovaries. Damaged cells are no longer able to perform their normal function.**

**Explain how damage to cells in the ovaries could result in an earlier onset of the menopause.**

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**[2]**

- (c) Women may become infertile several years before the menopause. Tests which predict the timing of the menopause would allow women to plan their pregnancies.

Recent genetic studies have identified *loci* which are associated with variation in the age of onset of natural menopause.

Each *locus* is a short DNA sequence which differs by one nucleotide.

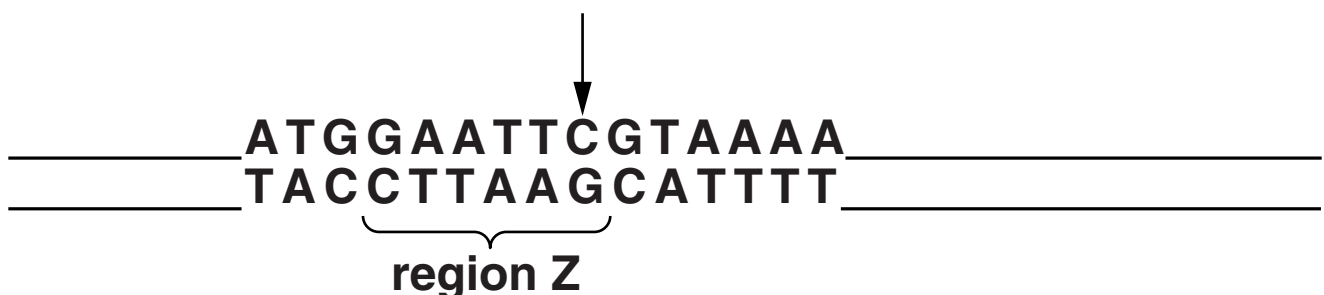
One variant increases the risk of an early menopause.

Fig. 6.1 is a diagram of a DNA sequence.

The variation occurs at the point indicated by the arrow where the nucleotide can be either 'C' or 'T'.

The presence of 'C' at this point increases the risk of an early menopause.

FIG. 6.1



- (i) What do the letters C and T represent in Fig. 6.1 opposite?

C \_\_\_\_\_

T \_\_\_\_\_

[1]

- (ii) Region Z is a nucleotide sequence that is complementary to the active site of an enzyme known as EcoRI. EcoRI is a type of enzyme that will cut the DNA strand.

State the following:

the type of enzyme that cuts DNA at a

particular recognition site \_\_\_\_\_

\_\_\_\_\_

the nature of the nucleotide sequences at the

recognition site \_\_\_\_\_

the name of the bond in the DNA strand that

the enzyme breaks \_\_\_\_\_

\_\_\_\_\_

[3]

- (iii) Separate DNA samples of the 'C' variant and the 'T' variant were treated using EcoRI. The DNA was analysed using electrophoresis.

Describe AND explain how the appearance of the pattern on an electrophoresis gel will differ between the DNA samples.

You should assume that:

region Z occurs only once

no other recognition sites for EcoRI are present.

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[3]

[TOTAL: 13]

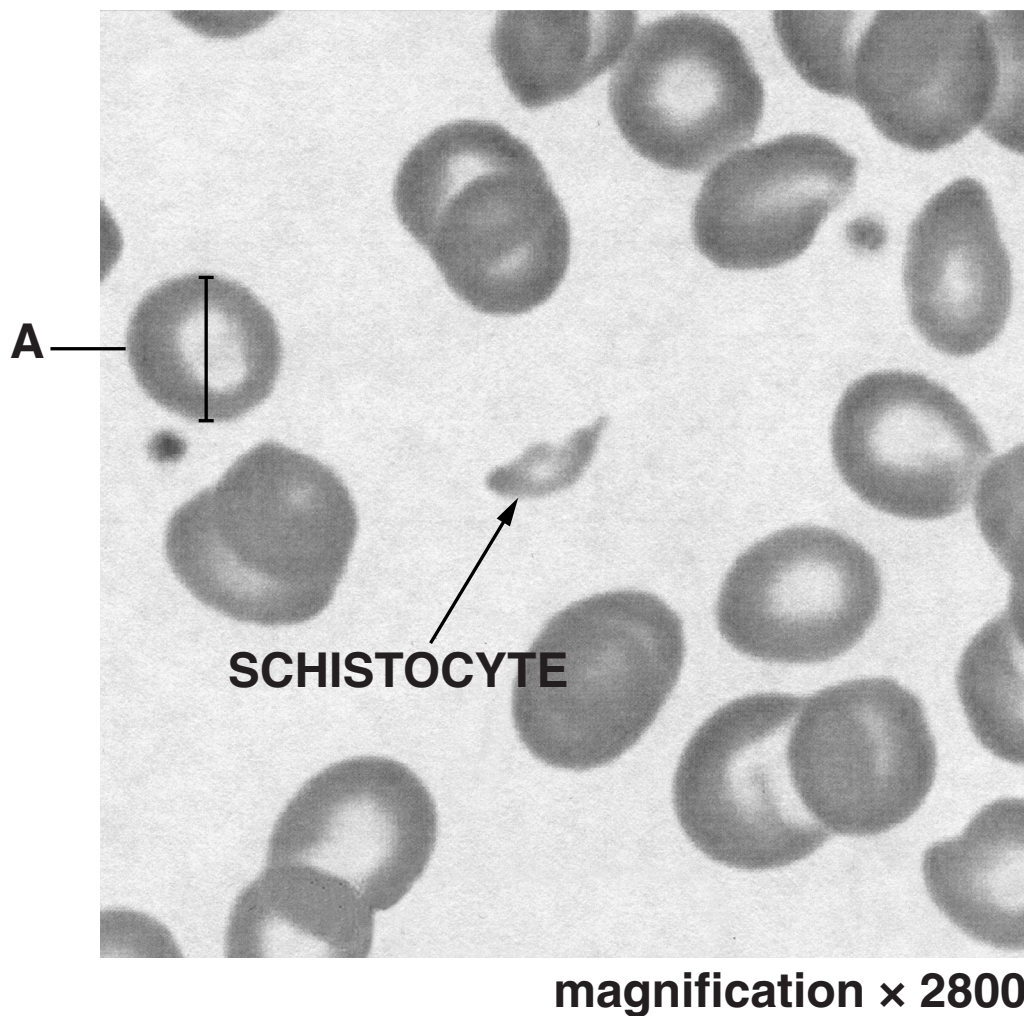


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- 7 The appearance of blood samples viewed under a microscope can be used to diagnose a number of different conditions.

Fig. 7.1 shows a blood sample. An abnormal cell known as a **SCHISTOCYTE** is labelled. Schistocytes are formed from fragments of red blood cells.

**FIG. 7.1**



- (a) (i) Use the information in Fig. 7.1 opposite to calculate the size of cell A.**

**Show your working. Give your answer to the nearest whole number.**

**Answer = \_\_\_\_\_  $\mu\text{m}$  [2]**

- (ii) When activated platelets bind to the endothelium of small arterioles and capillaries, schistocytes are formed.**

**Suggest why the binding of activated platelets leads to the formation of schistocytes.**

\_\_\_\_\_  
\_\_\_\_\_ [1]

- (iii) State one GENETIC disease that could be diagnosed by examination of a blood smear.**

\_\_\_\_\_ [1]

- (b) An infection by a strain of the bacterium *E. coli* called O157 can lead to the appearance of schistocytes in a blood sample.**

**Infections by this bacterium are the most common cause of acute kidney failure in children. This results in a condition called HUS.**

**Most children who develop HUS make a full recovery. A few children are left with permanent kidney damage.**

- (i) Name the short-term treatment given to children with acute kidney failure and OUTLINE the alternative long-term treatment options that would be available if necessary. [5]**

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- (ii) Suggest ONE further treatment that HUS patients may require to prevent them becoming anaemic. Explain your suggestion.**

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**[3]**

- (c) Some forms of HUS are not associated with *E. coli* O157 infections. These are known as atypical HUS or AHUS.

Some cases of AHUS are known to be due to an inherited gene mutation.

Carefully read and then complete the following passage about the inheritance of AHUS and the role of the genetic counsellor.

Some inherited forms of AHUS are due to a

\_\_\_\_\_ mutant allele.

Parents show no symptoms of the disease so, if

they have a child who develops AHUS, the child

must be \_\_\_\_\_ for this

allele. Since neither parent shows symptoms, yet

both carry the mutant allele, the allele does not

show \_\_\_\_\_ linkage.

**A genetic counsellor would use a**  
**\_\_\_\_\_ diagram to explain**  
**the risk of any further children either developing**  
**AHUS or being a carrier. Where unaffected parents**  
**have one child with AHUS, there would be a**  
**\_\_\_\_\_ percent chance of**  
**having a child who is a carrier. [5]**

**[TOTAL: 17]**

**END OF QUESTION PAPER**

**ADDITIONAL ANSWER SPACE**

**If additional answer space is required, you should use the following lined page(s). The question number(s) must be clearly shown in the margins.**




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