OCR
Oxford Cambridge and RSA

Tuesday 20 June 2017 – Morning

A2 GCE HUMAN BIOLOGY

F225/01 Genetics, Control and Ageing

Candidates answer on the Question Paper.

OCR supplied materials:
None

Other materials required:
- Electronic calculator
- Ruler (cm/mm)

Duration: 2 hours

Candidate forename		Candidate surname	

Centre number						Candidate number				

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(s) at the end of this booklet. The question number(s) must be clearly shown.
- Do **not** write in the barcodes.

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 The term 'colour blindness' is frequently used to describe a condition where an individual cannot distinguish between certain colours. This condition is more correctly described as a colour vision deficiency (CVD).

(a) (i) Outline how a test for CVD is performed.

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..... **[2]**

(ii) Name the part of the eye **and** the type of cell that is being assessed in a CVD test.

part of the eye .....

type of cell .....
[1]

(iii) CVD tests are designed to be carried out in a well-lit room.

Suggest why good lighting is essential for the validity of the CVD test.

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..... **[1]**

(b) The most common form of CVD is a red-green deficiency. This condition can be inherited as a sex-linked recessive allele.

Fig. 1 is a pedigree diagram for a family with a history of red-green deficiency.

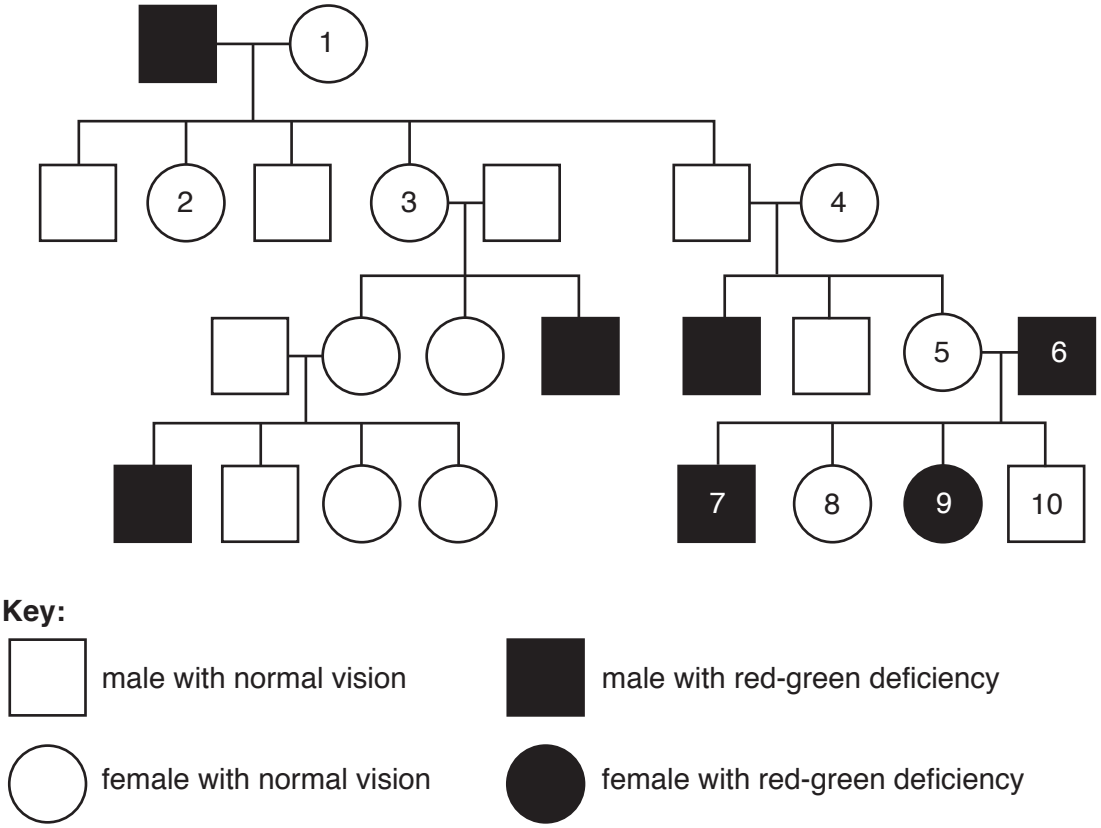

Fig. 1

Individual 1 has the genotype X^RX^R.

(i) What is the probability of individuals 2, 3 and 4 **all** being heterozygous for red-green deficiency?

Explain your answer.

probability = ...

explanation .....

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..... **[3]**

(ii) Construct a genetic diagram to explain the inheritance of the red-green deficiency allele in the children of individuals 5 and 6.

parent 5 genotype ..... parent 6 genotype .....

genotypes of children:

7 = ... 8 = ...

9 = ... 10 = ...

[3]

(iii) Suggest why genetic counsellors are rarely involved in families in cases of inherited colour vision deficiency.

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..... **[1]**

(c) The incidence of inherited red-green deficiency varies between populations.

Individuals with red-green deficiency:

- have difficulty in distinguishing between some ripe and unripe fruits
- have difficulty in distinguishing between meat that is raw or cooked.

In populations that rely more on hunting and gathering food, the incidence of inherited red-green deficiency is low.

Suggest why.

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..... **[3]**

(d) Colour vision deficiency can also be an acquired condition.

(i) One cause of acquired colour vision deficiency is the prolonged use of an antibiotic called **ethambutol**.

Suggest one disease where the length of the course of treatment could increase the risk of acquired colour vision deficiency.

Explain your suggestion.

disease .....

explanation .....

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..... **[2]**

(ii) Colour vision deficiency can also be a consequence of changes in the eye associated with ageing.

Name one age-related condition, **other than** macular degeneration, which could affect colour vision.

Suggest why this condition reduces colour vision.

condition .....

explanation .....

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..... **[2]**

[Total: 18]

2 One example of a eukaryotic organism that can be genetically modified to produce human proteins is the yeast *Saccharomyces cerevisiae*. Two advantages in using yeast cells as a source of human proteins are:

- Yeast cells secrete some of the proteins they produce into the cytoplasm.
- Yeast cells reproduce rapidly by a process known as budding.

(a) Fig. 2 is a diagram of a budding cell from *S. cerevisiae*.

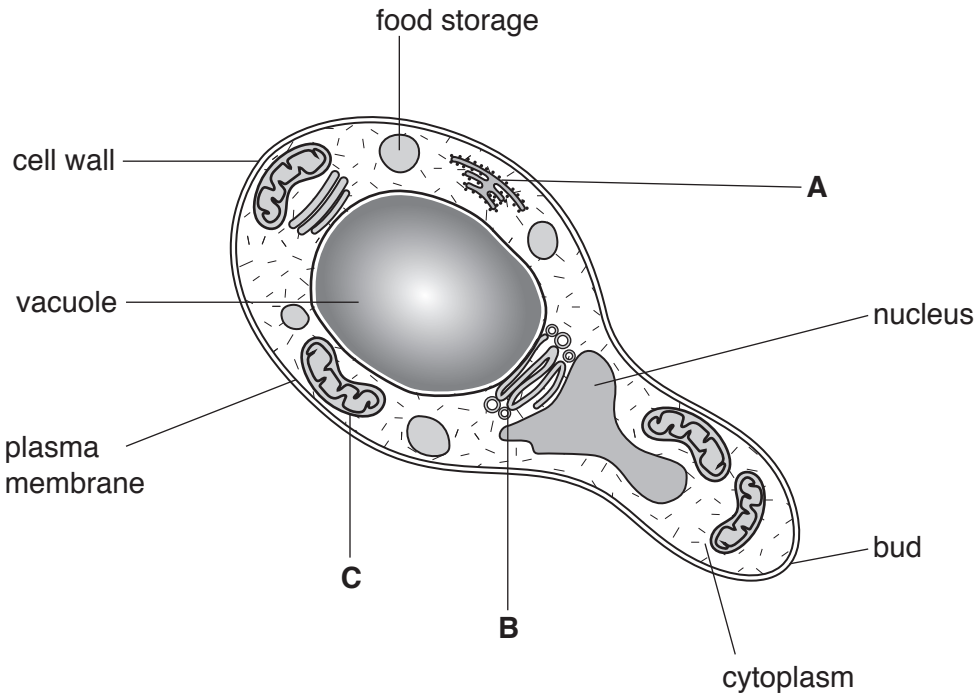

Fig. 2

(i) Identify the organelles **A**, **B** and **C** in Fig. 2 and explain how they are involved in the production and secretion of human proteins in a genetically modified yeast cell.

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..... **[4]**

(ii) Yeasts such as *S. cerevisiae* multiply by budding. The nucleus divides and the bud receives an identical copy of the cell's chromosomes.

Suggest **two** processes that must occur within the **nucleus** of *S. cerevisiae* before budding such as that shown in Fig. 2 can occur.

1 .....

2 .....

[2]

(iii) Chinese Hamster ovary cells are commonly used for genetic modification.

Using the information in Fig. 2, suggest why Chinese Hamster ovary cells are easier to genetically modify than yeast cells.

.....

..... **[1]**

(b) One advantage of using *S. cerevisiae* for genetic modification is its rapid growth rate.

Growth rates can be calculated by counting the yeast cells present in a culture.

(i) Name an instrument that could be used to count the number of yeast cells present in a culture.

..... **[1]**

(ii) Yeast cells have a generation time of **2 hours**.

Using the formula

$$\text{Log}(\mathbf{N}) = \mathbf{n}(\text{Log}2)$$

where **N** is the final number of yeast cells and

n is the number of generations,

calculate the time required for a single yeast cell to produce 100 000 cells.

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

Answer = ... hours **[2]**

(c) Like bacterial cells, *S. cerevisiae* cells also have plasmids.

Explain what you understand by the term plasmid and describe the role and use of plasmids in the genetic modification of **bacterial** cells.

In your answer you should refer to both the features of plasmids and their role in genetic modification.

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..... **[7]**

(d) Yeast cells that are URA3 negative ($URA3^-$) are used for genetic modification.

These cells do **not** have the gene URA3 in their genome and cannot grow in the absence of uracil.

URA3 codes for an enzyme necessary for yeast cells to synthesise uracil.

Plasmids used to genetically modify yeast cells contain the gene URA3.

(i) Suggest why the ability to synthesise uracil is essential for cell growth.

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..... **[2]**

(ii) What would you conclude had happened to $URA3^-$ yeast cells if they were **able** to grow in the absence of uracil?

..... **[1]**

[Total: 20]

3 Fig. 3 is a diagram of some of the structures and cells present in the pancreas.

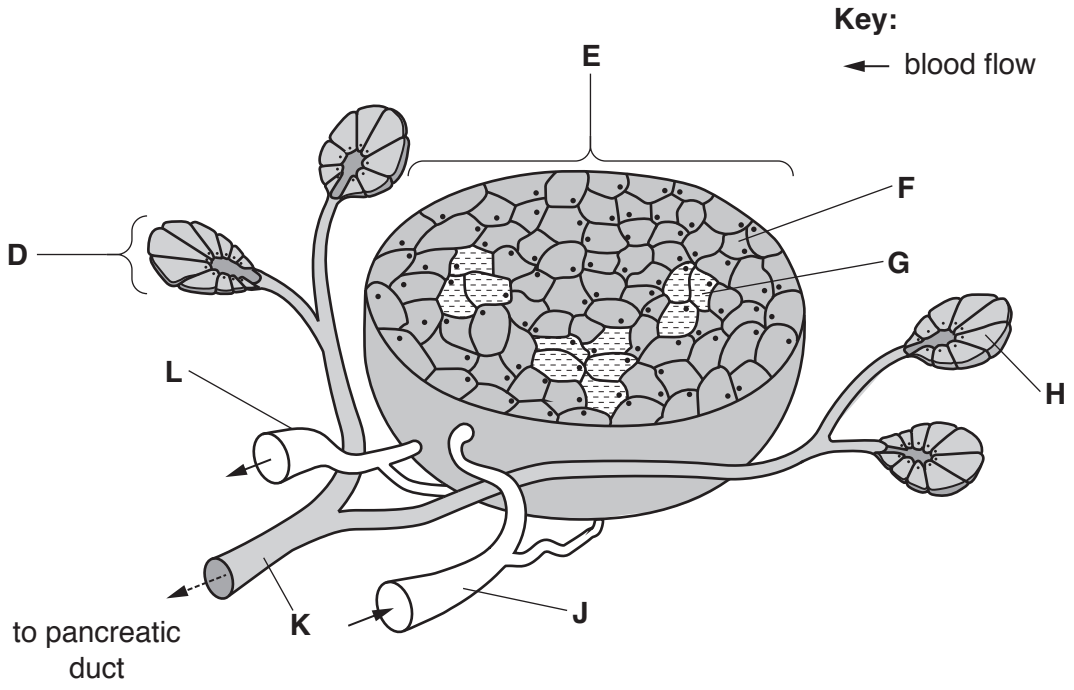

Fig. 3

(a) Using the information in Fig. 3, which letter or letters indicate:

(i) a type of cell that does **not** secrete a hormone?

letter(s) ..... **[1]**

(ii) an Islet of Langerhans?

letter(s) ..... **[1]**

(iii) a secretory cell?

letter(s) ..... **[1]**

(iv) a blood vessel linked directly to a venule?

letter(s) ..... **[1]**

(b) An investigation was carried out into the relationship between the presence of a virus in the lining of the intestine and type 1 diabetes.

Tests were carried out on a control group who were not diabetic and on a group known to have type 1 diabetes.

(i) Suggest how the investigators confirmed that the subjects with diabetes who were recruited to the trial had type 1 diabetes.

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..... **[2]**

Tissue samples from the lining of the intestine were taken and examined for the presence of virus particles or viral RNA.

The results from these tests are shown in Table 3.

	Response to tests for presence of virus (%)		
	Weak	**Moderate**	**Strong**
Patients with type 1 diabetes	23	41	10
Control subjects	24	5	0

Table 3

(ii) Using the information in Table 3, evaluate the evidence that links type 1 diabetes to a viral infection.

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..... **[3]**

(c) Type 1 diabetes has also been linked to the presence of certain antigens such as HLA-DR3 and HLA-DR4. HLA-DR antigens are part of the major histocompatibility (MHC) system.

Suggest why type 1 diabetes is more common in some families than others.

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..... **[1]**

(d) In the United Kingdom, approximately 3.5 million people have been diagnosed with diabetes. The majority of people diagnosed have type 2 diabetes.

(i) Outline the main risk factors associated with the **development** of type 2 diabetes.

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..... **[2]**

(ii) Explain why 3.5 million is an underestimate of the number of people with diabetes in the UK.

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..... **[2]**

[Total: 14]

4 Ageing is known to result in many changes to the cardiovascular system.

(a) The following paragraph describes some of the age-related changes that occur in cardiac muscle.

Complete the paragraph by inserting the most appropriate term.

There are several changes in cardiac muscle cells that occur as a result of ageing. The concentration of ..... ions in the cytoplasm does not decrease as rapidly following the onset of contraction. This is thought to be due to a decrease in the activity of the ATP-driven pump in the membrane of the ..... .

As a consequence the ventricle walls contract for longer. This means less time is available for the ventricles to fill as the length of ..... is shorter.

In addition, the binding of the neurotransmitter noradrenaline no longer produces an effect. This transmitter is released by sympathetic ..... neurones in the autonomic nervous system. This means that the heart rate will not ..... in response to a fall in blood pressure. This breakdown in negative feedback is one aspect of ..... which declines with age.

[6]

(b) Ageing also produces changes in the functioning of blood vessels.

- The **compliance** of an artery is a measure of the ability of the artery wall to stretch in response to an increase in blood volume.
- There is a negative correlation between compliance and blood pressure.

Fig. 4 shows the relationship between age and compliance in two groups:
- sedentary subjects who exercised very little
- endurance trained athletes who exercised frequently.

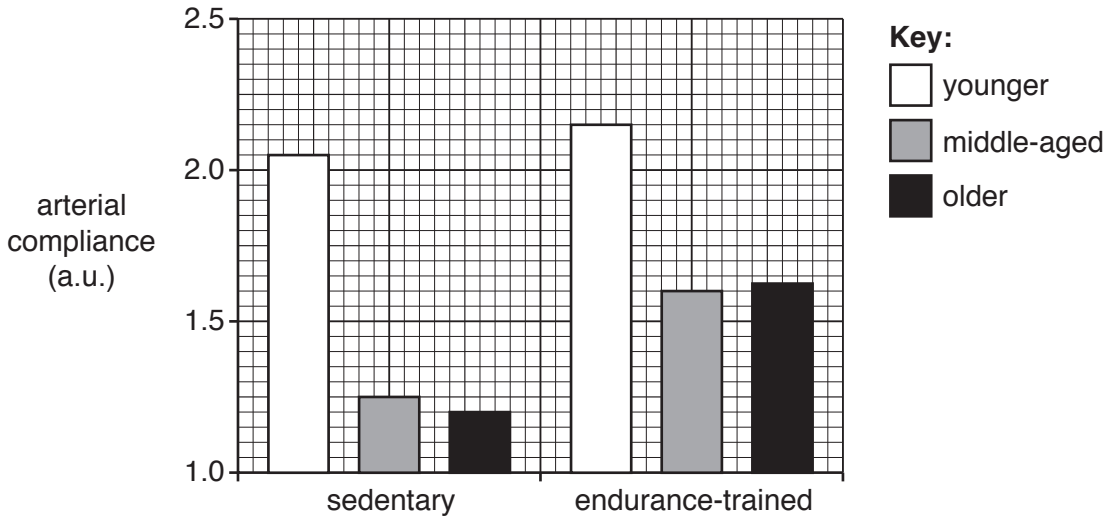

Fig. 4

A student wrote the following statement about the data in Fig. 4.

'These data explain why the risk of coronary heart disease increases with age and that exercising will significantly reduce that risk.'

Explain whether or not the data in Fig. 4 support the statement made by the student.

In your answer you should use data and explain the link between compliance and coronary heart disease.

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..... **[7]**

(c) Arterial compliance is also known to decrease following the menopause. Medication taken to relieve the symptoms of the menopause has been shown to improve arterial compliance.

(i) Suggest one form of treatment for menopausal symptoms **other than HRT** that could improve arterial compliance.

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..... **[1]**

(ii) Describe **two** additional physiological changes associated with the menopause.

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..... **[2]**

[Total: 16]

5 The kidneys are organs responsible for the removal of urea and for the maintenance of water potential in blood plasma.

(a) Explain why urea is correctly described as an excretory product.

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..... **[2]**

Question 5(b) begins on page 18

(b) Fig. 5.1 shows the relationship between the **osmolality** of blood plasma and the concentration of ADH present in the plasma.

- The osmolality of a solution is a measure of the concentration of the solutes affecting the water potential of that solution.
- Osmolality is expressed as milliosmoles per kilogram (mosm kg^{-1}).

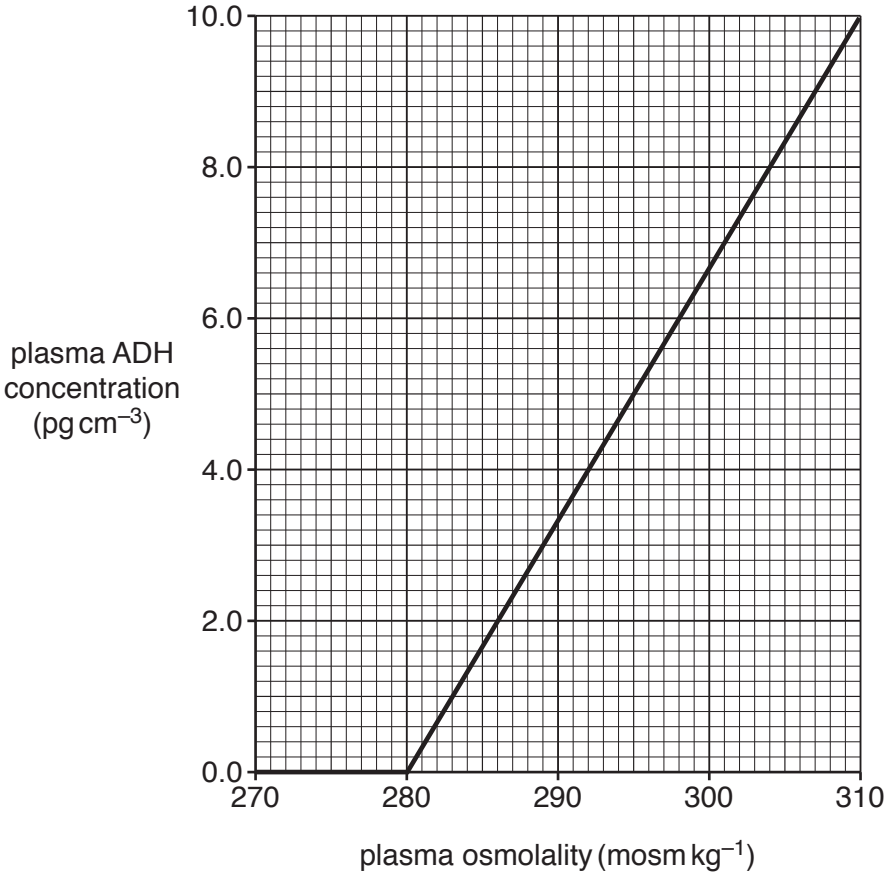

Fig. 5.1

(i) Describe the events in the body that result in an increase in the concentration of plasma ADH when plasma osmolality increases above 280 mosm kg^{-1}.

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..... **[4]**

(ii) On Fig. 5.2, complete the graph to show the relationship between plasma concentrations of ADH and the osmolality of **urine**.

Fig. 5.2

[2]

(c) Several different diseases can result in kidneys becoming damaged. When kidney damage is severe, a number of different treatments are available to patients.

Fig. 5.3 shows a patient with kidney damage undergoing one form of treatment.

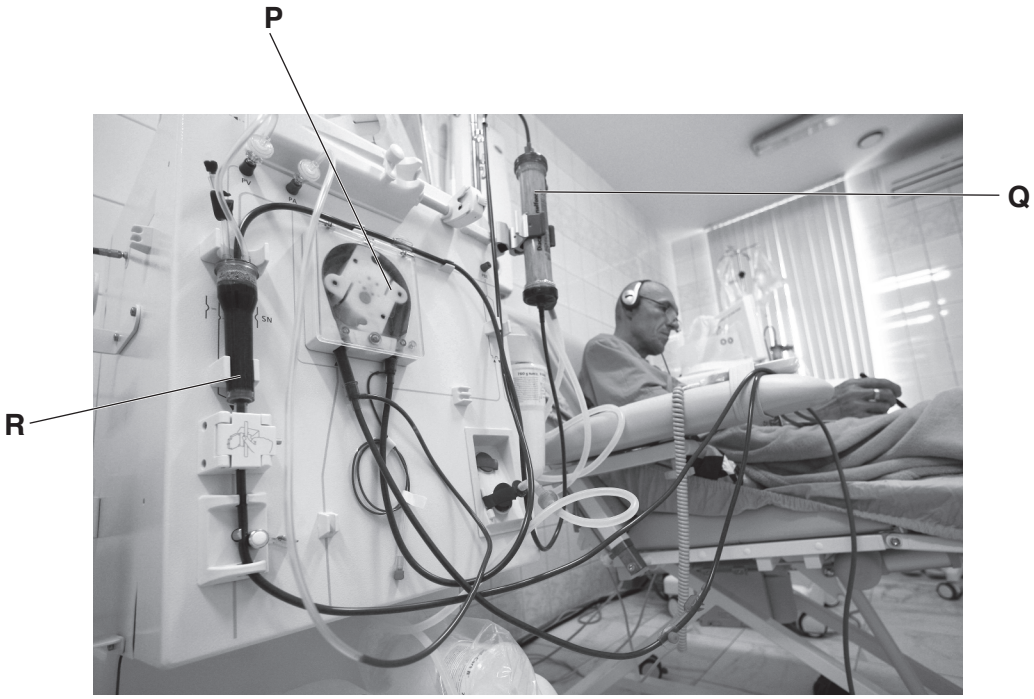

Fig. 5.3

(i) Identify the type of treatment shown in Fig. 5.3.

..... **[1]**

(ii) State the role of the part of the apparatus labelled **P**.

..... **[1]**

(iii) In the part of the apparatus labelled **Q**, blood is flowing through tubes made from an artificial membrane. The tubes are surrounded by a fluid.

State one property of this artificial membrane that is essential to the functioning of **Q**.

..... **[1]**

(iv) Blood flows through the part of the apparatus labelled **R** before finally being returned to the patient.

What is the function of **R**?

..... **[1]**

(d) In the future, new techniques such as that shown in Fig. 5.4 could be used to produce replacements for damaged organs such as kidneys.

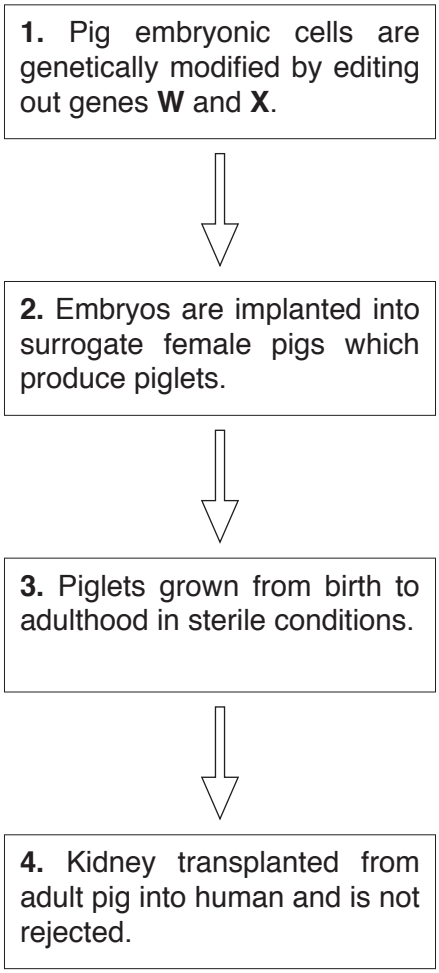

Fig. 5.4

The following descriptions refer to the stages in the technique shown in Fig. 5.4.

State the term or terms which corresponds to each of the following descriptions:

(i) The type of molecules coded for by genes **W** and **X** in stage **1**.

..... **[1]**

(ii) The precise site of implantation of the modified pig embryos in stage **2**.

..... **[1]**

(iii) The type of cell division happening in stage **3**.

..... **[1]**

(iv) The type of transplantation described in stage **4**.

..... **[1]**

(e) Techniques such as that described in Fig. 5.4 raise a number of ethical issues.

(i) Identify **one** ethical issue associated with each of the following stages:

stage 3 .....

.....

stage 4 .....

.....

[2]

(ii) Suggest how you could justify the use of techniques such as that described in Fig. 5.4.

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..... **[2]**

[Total: 20]

6 In the UK, about 15% of strokes are caused by bleeding in or around the brain.

- A bleed in the brain is referred to as an intracerebral haemorrhage.
- Blood leaks into the surrounding area and results in the death of brain cells.
- Injury is the most common cause of intracerebral haemorrhage in people aged below 50.

(a) (i) What term is used to describe an intracerebral haemorrhage which occurs as a result of injury?

..... **[1]**

(ii) Name the protein present in blood that inhibits the repair of neurones in the CNS **and** describe the role of this protein in limiting further damage in the brain following a bleed.

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..... **[4]**

(iii) Suggest one therapeutic drug that may increase the risk of an intracerebral haemorrhage.

..... **[1]**

(b) Following a stroke, patients frequently receive visits from health professionals such as occupational therapists.

Fig. 6 is an extract from some of the notes made by an occupational therapist while visiting a stroke patient.

9:30 am	Arrived and admitted by client's husband. She was surprised to see me even though I had phoned at 9:00 to tell her I was on my way. She asked my name and seemed happy to sit and talk and offered me tea which I refused. Proceeded with my assessment.
9.40 am	Client asked my name and again offered me tea. .....
11:00 am	Completed my assessment. Client accompanied me to the door and asked me to write down my name and when I was coming again on her calendar.

Fig. 6

(i) Using the information in Fig. 6, what evidence suggests that there has been a loss of short term memory?

.....

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..... **[1]**

(ii) Suggest one technique **other than** the use of notes or a calendar that can be used to improve memory in stroke patients.

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..... **[1]**

(c) Approximately 85% of strokes are caused by a blockage in an artery leading to the brain which interrupts or limits the flow of blood to brain tissue. These are known as **ischaemic** strokes.

Name a technique that could be used to distinguish between an ischaemic stroke and a haemorrhagic stroke.

..... **[1]**

(d) A study was carried out on the possible link between the use of cannabis and the risk of an ischaemic stroke.

- A urine test was carried out on 160 stroke patients and on a control group.
- The control group was matched for age and gender.
- A positive test for cannabis was recorded in 8% of the control group.

(i) A positive test for cannabis was recorded for 16% of the stroke patients.

Calculate the number of stroke patients who tested positive for cannabis use.

Show your working.

Answer ..... **[2]**

(ii) The researchers reached the following conclusion:

The study provides evidence of an association between cannabis and stroke ..... but the association is confounded because all but one of the cannabis users also used tobacco regularly.

Suggest why the use of tobacco makes it difficult to draw a conclusion about the link between cannabis use and strokes.

.....

..... **[1]**

[Total: 12]

END OF QUESTION PAPER

ADDITIONAL ANSWER SPACE

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