

**Friday 20 June 2014 – 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**




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| Candidate<br>forename |  | Candidate<br>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 pages 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 **32** pages. Any blank pages are indicated.

Answer **all** the questions.

1 MRI scans have been used to investigate the structure and function of the human brain.

Scans are essential when health professionals suspect that a stroke has occurred.

Fig. 1.1 is an MRI scan of the brain.



**Fig. 1.1**

(a) Identify the following structures labelled on Fig. 1.1.

| Letter | Name of structure |
|--------|-------------------|
| B      |                   |
| C      |                   |

[2]

(b) Mrs G, a 72-year-old pensioner, collapsed while out shopping and an ambulance was called. The paramedics suspected a stroke and this was later confirmed at the hospital by a doctor. The doctor in Accident and Emergency based his diagnosis on a number of observations including the following:

- non-normal response to a reflex test on the foot (the plantar reflex)
- muscular weakness, decreased touch sensation and reduced awareness on the right side of the body
- mild confusion and slurring of speech.

(i) State what is meant by a 'reflex test' **and** give **one** example of a reflex test other than the plantar reflex.

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(ii) Using the doctor's observations, suggest which part of the brain has been affected by the stroke.

..... [1]

(c) Following an MRI scan, Mrs G was found to have an area of her brain that was deprived of oxygen due to a blood clot. She was immediately 'thrombolysed' and then transferred to a specialist stroke ward.

- 'Thrombolysis' involves the injection of a substance known as a plasminogen activator.
- The activator converts the protein **plasminogen** in blood into an active form called **plasmin**.
- Plasmin dissolves blood clots.

(i) Suggest the type of molecule that plasmin might be **and** how it dissolves blood clots.

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..... [2]

- (ii) Plasminogen activator is a glycoprotein. The plasminogen activator used in hospitals has been manufactured using genetically modified cells.

What **type** of modified cell might be used in the manufacture of plasminogen activator?

Explain your answer.

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..... [2]

- (iii) The recommended dose of plasminogen activator is 0.9mg for each kilogram of body mass.

- Mrs G was prescribed a dose of 67.5 mg.
- Mrs G is 1.5m tall.
- BMI is calculated using the formula below:

$$\frac{\text{body mass (kg)}}{(\text{height (m)})^2}$$

Using the above information, calculate a BMI for Mrs G.

Give your answer to the nearest whole number.

Answer = ..... [2]

- (iv) Using your calculation of BMI in part (iii), discuss the risk factors that may have contributed to Mrs G's stroke.

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..... [2]

- (d) Following several days on the specialist stroke ward, Mrs G was recovering well but appeared to have some problems with her short-term memory.

She was transferred to a rehabilitation unit where health professionals started a programme to improve her independence.

- (i) Outline what is meant by 'short-term memory loss'.

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..... [1]

- (ii) Suggest **two** different techniques that may be used to improve short-term memory in stroke patients.

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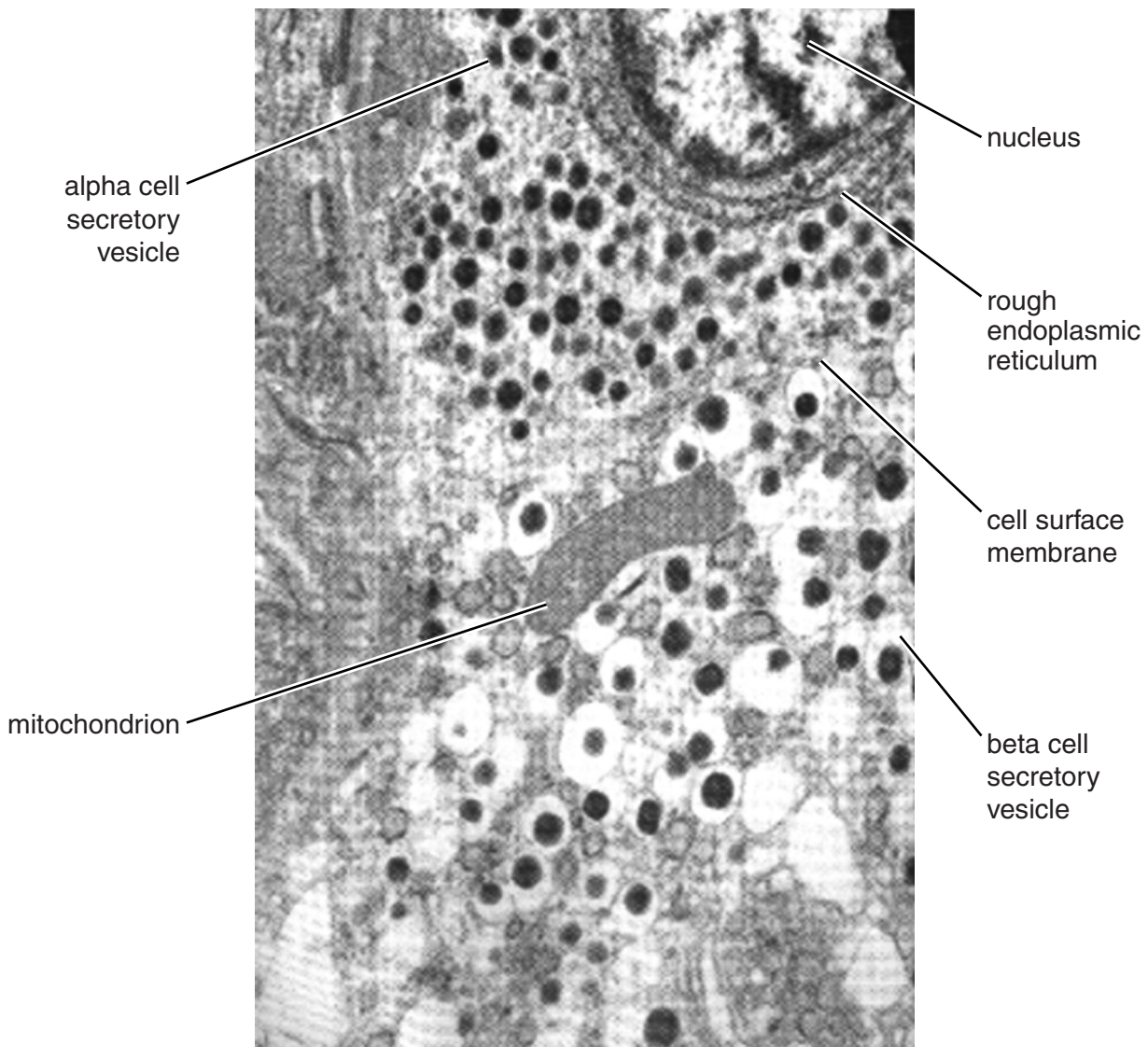
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[Total: 17]

2 Recent studies, such as those carried out by the World Health Organisation, have noted an increase in the global incidence of **Type 1** diabetes.

(a) Type 1 diabetes occurs due to changes in pancreatic tissue. Fig. 2.1 shows a photomicrograph taken of a section through pancreatic tissue.



**Fig. 2.1**

(i) State the region of the pancreas from which this tissue section has been taken.

..... [1]

(ii) Which of the secretory vesicles contains the hormone that lowers blood glucose?

..... [1]

(b) The hormones produced by the region of the pancreas shown in Fig. 2.1 are both polypeptides.

Explain how these hormones are:

- produced by the secretory cells
- released from the secretory cells
- transported to their target organs.



*In your answer you should refer to the organelles shown in the diagram and identify the target organs.*

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**Answer lines for this question continue on page 8**

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[Total: 10]



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**Question 3 begins on page 10**

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3 Human genetic diseases, such as sickle cell anaemia and cystic fibrosis, are caused when mutated forms of a gene are inherited.

The study of how these alleles are distributed in human populations can be an indication of how a human population has evolved over time.

(a) State **two** genetic diseases, other than sickle cell anaemia and cystic fibrosis, which are caused by the inheritance of mutant alleles.

1 .....

2 .....

[2]

(b) The frequency of the sickle cell allele in human populations has been found to be higher in areas of the world such as India and the west coast of Africa. In these regions, malaria is said to be endemic.

(i) State what is meant by the term *endemic*.

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(ii) Explain why the frequency of the sickle cell allele is higher in areas such as India and the west coast of Africa.

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- (c) The mutation responsible for cystic fibrosis occurs in a gene coding for a membrane transport protein called CFTR.

In March 2012, a gene therapy trial for the treatment of cystic fibrosis was launched in the United Kingdom. Patients received the treatment by inhaling small liposomes (phospholipid vesicles) containing molecules of DNA with a copy of the normal CFTR gene. The liposomes act as vectors and will deliver this DNA into the cells lining the lungs.

- (i) Explain why a treatment such as that described above is an example of somatic gene therapy.

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- (ii) Suggest why it was necessary to enclose the DNA in liposomes for delivery into cells.

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- (d) The CFTR gene is found on chromosome 7 and is approximately 190 000 base pairs in length.

The copy of the normal CFTR gene was made using an enzyme called **reverse transcriptase**.

- Messenger RNA coding for the CFTR protein is isolated from cells.
- Reverse transcriptase is used to synthesise a DNA strand that is complementary to the mRNA molecule.
- The DNA molecule is made double stranded forming a complementary or **cDNA** molecule containing a copy of the gene.
- ‘Sticky ends’ are added to this cDNA molecule.
- This DNA is inserted into a plasmid to form a recombinant plasmid.

- (i) Suggest why a cDNA copy of the CFTR gene will have fewer base pairs than the CFTR gene located on chromosome 7.

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..... [2]



(e) The introduction of normal, functional copies of the CFTR gene into cells allows the cells to produce temporarily a functioning protein.

- The CFTR gene is **not** inserted into the genome of the cells.
- To maintain the production of the protein, the gene needs to be introduced into the cells lining the lungs over and over again.
- This means that repeated doses of gene therapy using fat globule vectors are required.

Suggest why the failure of the CFTR gene to incorporate into the cell genome means that the beneficial effect of the gene therapy is only temporary.

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(f) Viruses can also be used as vectors.

- Research is being carried out into the use of modified viruses to deliver the CFTR gene into cells.
- Viruses have been shown to be very successful vectors for gene therapy.
- Treatments using most viruses become less and less successful at delivering the gene into cells with each repeated treatment.

Suggest why, with repeated treatments, viruses become less and less successful as vectors of DNA into cells.

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[Total: 22]

14  
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4 Age-related macular degeneration (AMD) is one of the leading causes of irreversible sight loss in developed countries such as the United Kingdom.

AMD results in changes in the retina leading to the loss of visual acuity.

(a) (i) State what is meant by *visual acuity* and describe briefly how visual acuity is assessed.

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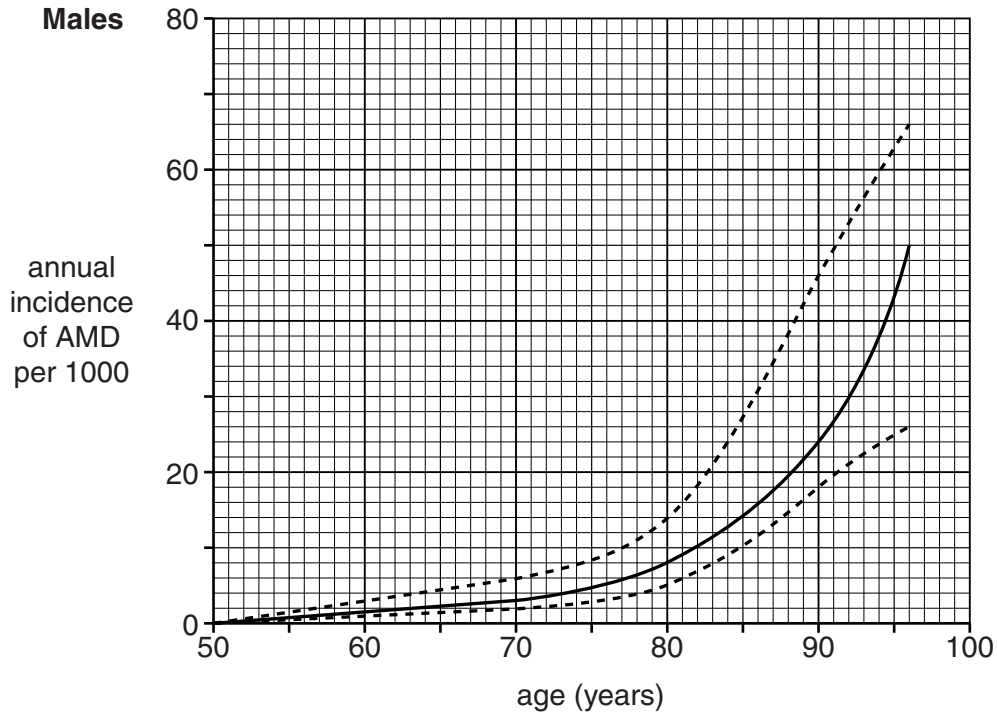
(ii) Explain why damage to the macula leads to loss of visual acuity.

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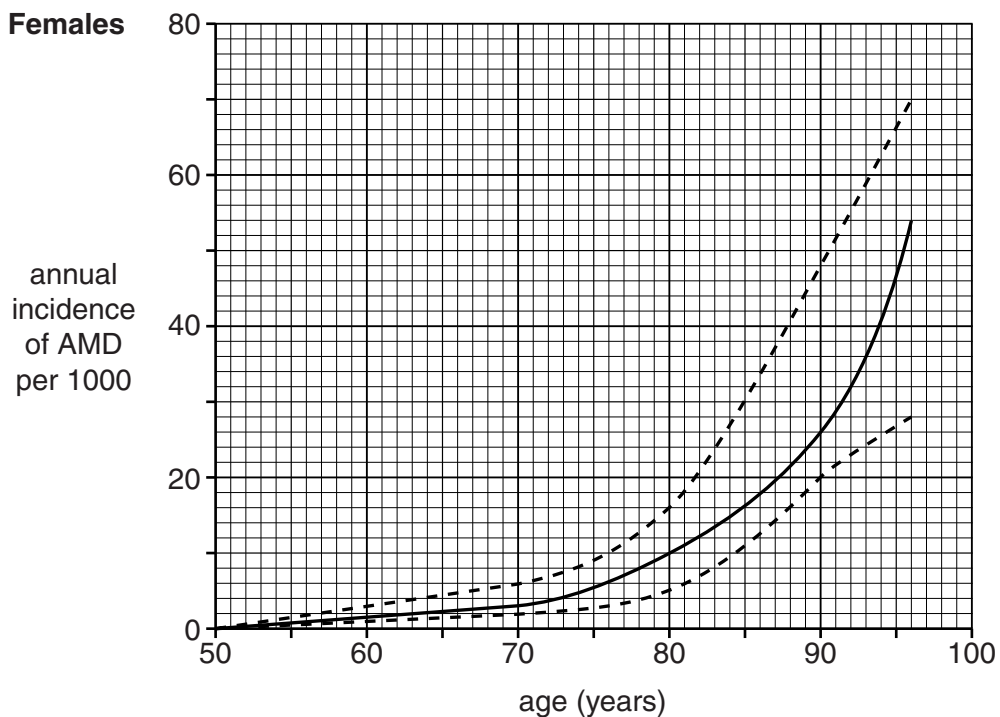
- (b) Studies have been carried out to estimate the incidence of AMD within the population of **any** developed country in **any** given year.

Figs. 4.1(a) and 4.1(b) show the estimated changes in incidence of AMD with age as solid lines. The 95% credible intervals for the data are shown by dotted lines. A credible interval represents the range of values within which the true value is expected to lie.

Fig. 4.1(a) shows the estimated change in incidence in males and Fig. 4.1(b) shows the estimated change in incidence in females.



**Fig. 4.1(a)**



**Fig. 4.1(b)**



(i) What is indicated by the 95% credible interval?

Insert a tick (✓) against the **most** appropriate response in the table below.

| Response                                  | Insert a tick (✓) |
|---|-------------------|
| The data are 95% accurate                 |                   |
| The validity of the data is 95%           |                   |
| The confidence limits of the data are 95% |                   |

[1]

(ii) Compare the changes in incidence of AMD with age in males and females **and** comment on the reliability of the estimates.

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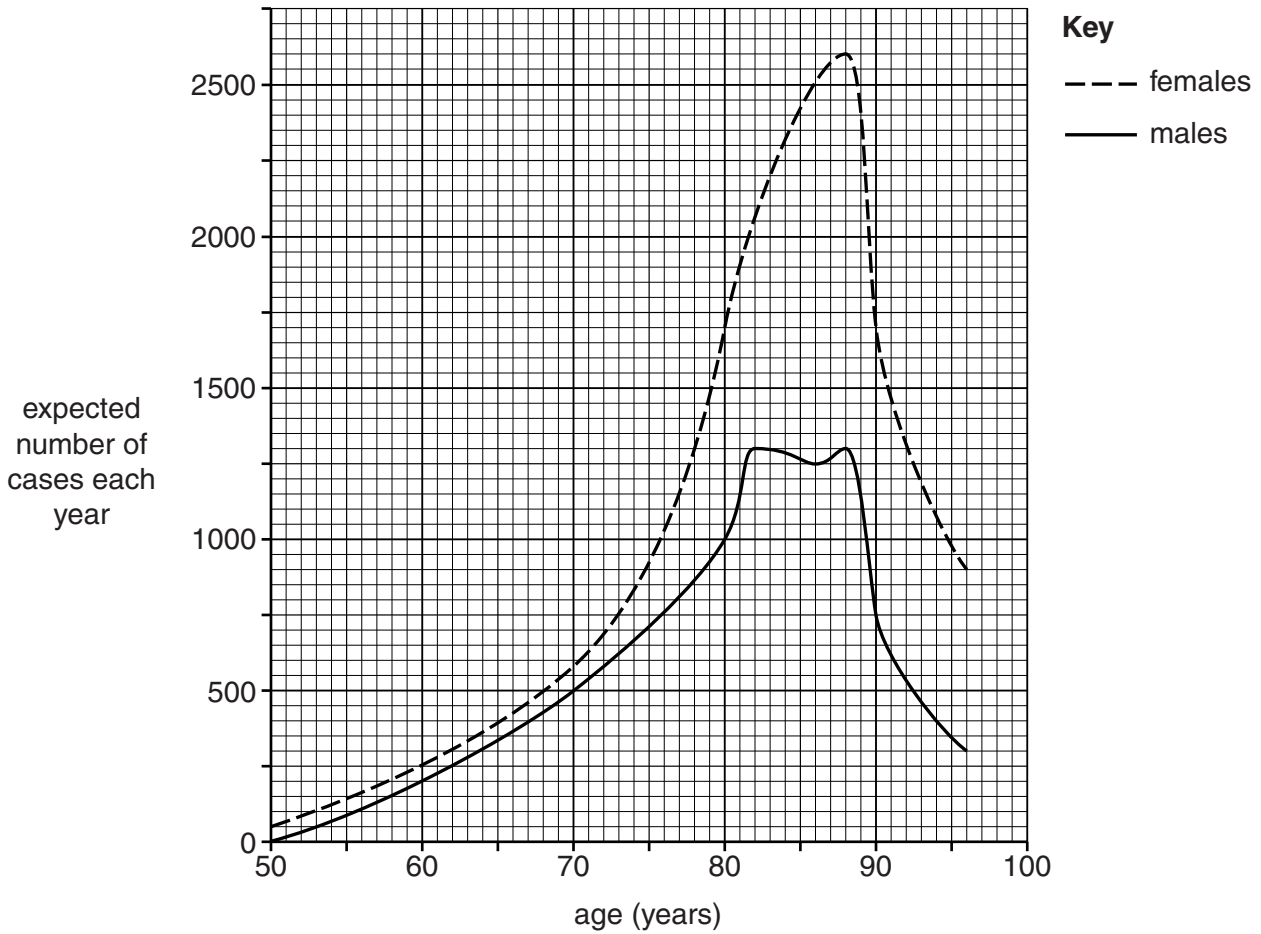
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(c) Fig. 4.2 shows the changes in the **expected number of cases** of AMD with age if the **estimates** of incidence (Fig. 4.1(a) and Fig. 4.1(b)) are applied to the United Kingdom's population.



**Fig. 4.2**

(i) Suggest what additional information would be required to convert the dependent variable in Fig. 4.1(a) and Fig. 4.1(b) to the dependent variable in Fig. 4.2.

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 ..... [1]

(ii) Explain why the **pattern** shown by the data in Fig. 4.1(a) and Fig. 4.1(b) is different to that shown in Fig. 4.2.

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 ..... [2]

(d) One form of AMD, known as **neovascular** AMD or **wet** AMD, is caused by the growth of blood vessels into the macula.

- A growth factor is normally released by cells in response to low oxygen concentrations.
- The growth factor stimulates the growth of blood vessels.
- Blood vessels may leak causing very rapid loss of vision.

(i) Suggest **one** factor, other than ageing, which may increase the risk of developing wet AMD.

Explain your suggestion.

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..... [2]

(ii) The growth of blood vessels is stimulated by a growth factor called **VEGF**.

**VEGF** binds to receptors on the cell surface membrane of endothelial cells.

One treatment available for wet AMD involves blocking the action of **VEGF** using a modified antibody.

Suggest how the modified antibody prevents the action of **VEGF**.

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(e) Fig. 4.3 shows the results from one clinical trial of the modified antibody treatment.

At 3-monthly intervals, each patient’s visual acuity was measured and compared to their visual acuity at the beginning of the trial. A positive value indicates an improvement and a negative value indicates a decline.

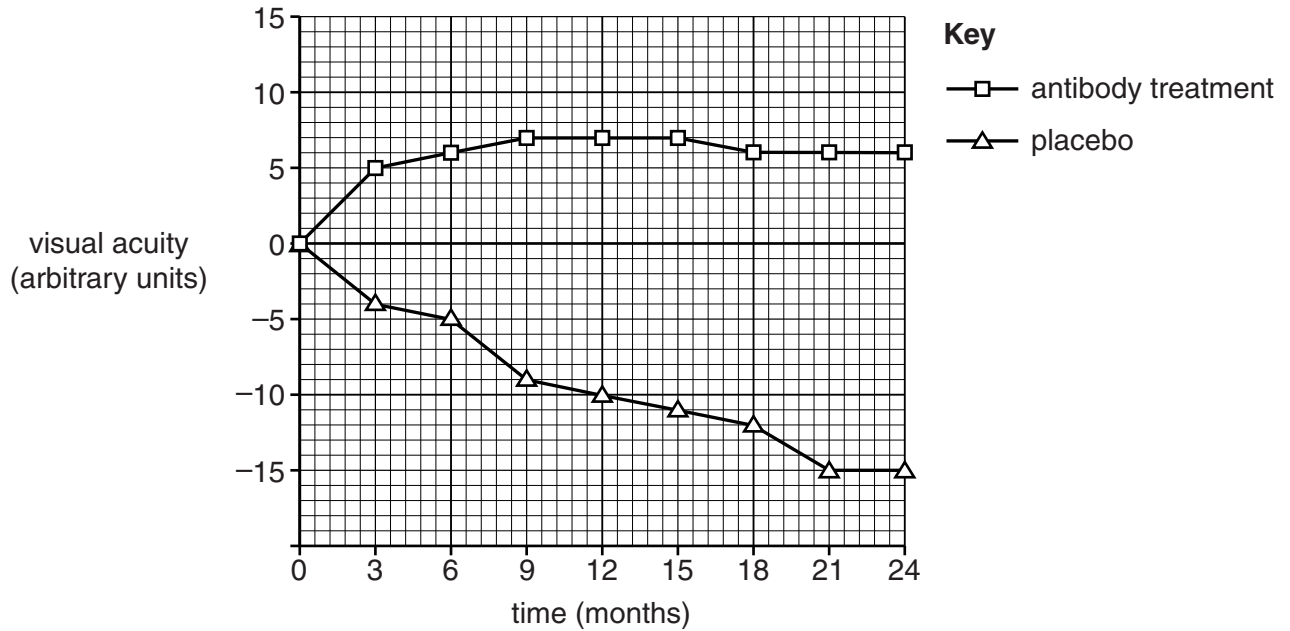


Fig. 4.3

(i) State what may be concluded about the effectiveness of the modified antibody treatment based on the results of this clinical trial.

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(ii) Suggest **one** further piece of information that would be required in order for your conclusion to be valid.

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..... [1]

[Total: 20]

**21**  
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**Question 5 begins on page 22**

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- 5 In humans, the kidneys are a key organ in the excretory system. They are also essential for the maintenance of blood volume and electrolyte balance.

Changes in the functioning of the kidneys can be detected by monitoring the composition of urine.

- (a) Explain why the kidneys can be considered as **excretory organs**.

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- (b) The functional unit of the kidney is the nephron. Fig. 5.1 shows a simplified diagram of a nephron.

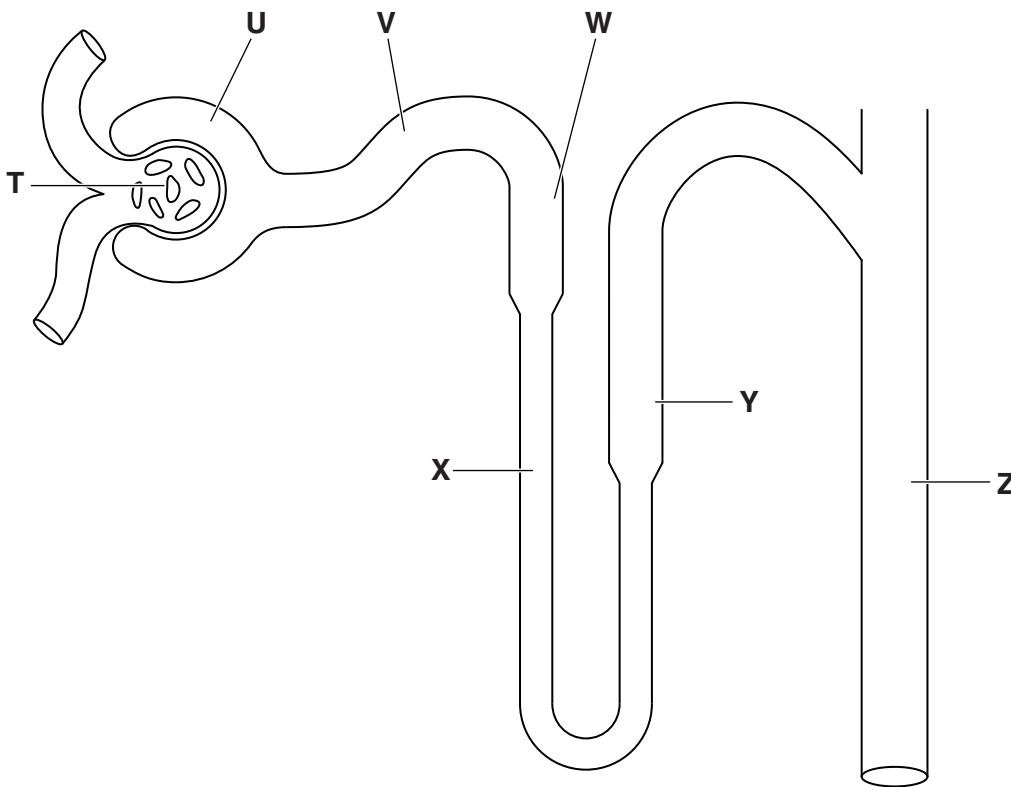


Fig. 5.1

- (i) Identify the structures **T** and **U** and state the region of the kidney where these structures are found.

**T** .....

**U** .....

Region of kidney .....

[3]

- (ii) Complete the following paragraphs about the passage of filtrate through the loop of Henle by inserting the most appropriate word.

Electrolytes such as sodium ..... are actively reabsorbed from the filtrate at region **Y**, which is the ..... limb of the loop of Henle. This reabsorption reduces the ..... of the surrounding tissues in the ..... of the kidney.

The walls of the tubule in region **X** are ..... to water. The result is that water is reabsorbed from the filtrate by the process of .....

[6]

- (c) Reabsorption of glucose in the nephron takes place actively using glucose transport proteins. These transport proteins are located in regions **V** and **W** of the nephron.

In a condition known as familial glycosuria, the glucose transport proteins in region **W** are not functioning. This results in glucose being present in the urine.

- (i) Predict the effect of a non-functioning glucose transport protein on the **volume** of filtrate leaving the loop of Henle. Explain the reason for your prediction.

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- (ii) Suggest why the management of familial glycosuria may involve a genetic counsellor.

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[Total: 15]

6 Ageing can be a very different experience for a person depending on where in the world they live. One consequence of ageing that many people experience is **osteoarthritis**.

(a) Distinguish between osteoarthritis and osteoporosis.

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..... [2]

(b) The World Health Organisation collects data on the global distribution of many diseases including **osteoarthritis**. The statistics that are calculated are used in a range of epidemiological studies.

One important statistic that can be calculated is the disability-adjusted life year or **DALY**.

- One DALY can be considered as one lost year of **healthy** life.
- In North America the DALY for osteoarthritis is 796 000.
- In Africa the DALY for osteoarthritis is 650 000.

Another important statistic that can be calculated is life expectancy at birth.

- In North America life expectancy is 74.
- In Africa life expectancy is 50.

Both sets of statistics are based on data collected in the year 2000 and are for both males and females.

(i) How does the statistical evidence support the conclusion that the onset of osteoarthritis in Africa happens at an earlier age?

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..... [2]



(ii) Suggest why the onset of osteoarthritis happens at an earlier age in Africa.

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(iii) The data used for calculating the DALYs for osteoarthritis were combined data from males and females.

Explain why it would **not** be appropriate to combine male and female data for epidemiological studies on **osteoporosis**.

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[Total: 8]

Question 7 begins on page 26

7 Many researchers are involved in investigating the potential use of stem cells to treat a variety of human conditions.

Stem cells can be produced from a variety of sources. One possible source is known as somatic cell nuclear transfer (**SCNT**).

In **SCNT**:

- the nucleus is removed from a donor’s oocyte
- a nucleus from the patient’s somatic cell is introduced into the oocyte
- the resultant cell is allowed to divide to produce several cells
- some of these cells will be used to produce cultures of stem cells.

(a) Insert a tick (✓) against the term that best describes the production of stem cells using SCNT.

| Term                 | Insert a tick (✓) |
|----------------------|-------------------|
| Genetic engineering  |                   |
| Therapeutic cloning  |                   |
| Reproductive cloning |                   |
| ICSI                 |                   |
| IVF                  |                   |

[1]

(b) Stem cells have the potential to be used to treat patients with conditions such as Parkinson’s disease or Type 1 diabetes.

(i) Suggest the **type** of stem cell which is produced using SCNT.

..... [1]

(ii) State **two** properties of the stem cells produced by SCNT that would make them potentially suitable for treating conditions such as Parkinson's disease **or** Type 1 diabetes.

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[2]

[Total: 4]

Question 8 begins on page 28

8 Hot flushes are one of the symptoms associated with the menopause. These symptoms are thought to be linked to falling oestrogen levels.

(a) Low levels of oestrogen are thought to disrupt the thermoregulatory centre in the brain.

Name the part of the nervous system which includes the brain **and** state the exact location of the thermoregulatory centre.

Part of nervous system .....

Exact location .....

[1]

(b) Hot flushes may last from 3 to 10 minutes.

Scientists investigating hot flushes carried out the following measurements to monitor changes in the body during a hot flush:

- blood flow in the skin
- sweating
- heart rate.

At the onset of a hot flush, there is a sudden increase in sweating. This is followed by vasodilation, which causes an increase in blood flow in the skin. Shortly after this, there is an increase in heart rate.

(i) Suggest why an increase in blood flow in the skin might result in an increase in heart rate.

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..... [1]

(ii) Suggest **one further** effect on the body of increased vasodilation and sweating.

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..... [1]

- (c) Symptoms of the menopause such as hot flushes can be treated by hormone replacement therapy (HRT).

Some forms of HRT are provided as implants.

Explain why cyclical HRT treatments are not available as implants.

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..... [1]

[Total: 4]

**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.

A large area of lined paper for writing answers. It features a vertical margin line on the left side and horizontal dotted lines for writing. The lines are evenly spaced and extend across the width of the page.



A large area of the page is filled with horizontal dotted lines, providing a space for writing answers. A solid vertical line runs down the left side of this area, creating a margin.



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