THIS IS A NEW SPECIFICATION

OCR
RECOGNISING ACHIEVEMENT

ADVANCED GCE

HUMAN BIOLOGY

Genetics, Control and Ageing

F225

Candidates answer on the Question Paper

OCR Supplied Materials:
None

Other Materials Required:
- Electronic calculator
- Ruler (cm/mm)

Wednesday 16 June 2010
Morning

Duration: 1 hour 45 minutes

OCE/14310

Candidate Forename		Candidate Surname	

Centre Number						Candidate Number				

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. Additional paper may be used if necessary but you must clearly show your Candidate Number, Centre Number and question number(s).

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**.
- You are advised to show all the steps in any calculations.
- Where you see this icon you will be awarded marks for the quality of written communication in your answer.
- This document consists of **24** pages. Any blank pages are indicated.

Answer **all** the questions.

1 The kidney is responsible for the removal of nitrogenous waste from blood plasma.

Fig. 1.1 shows a photomicrograph of a section through the cortex of the kidney and a simplified drawing of some of the structures.

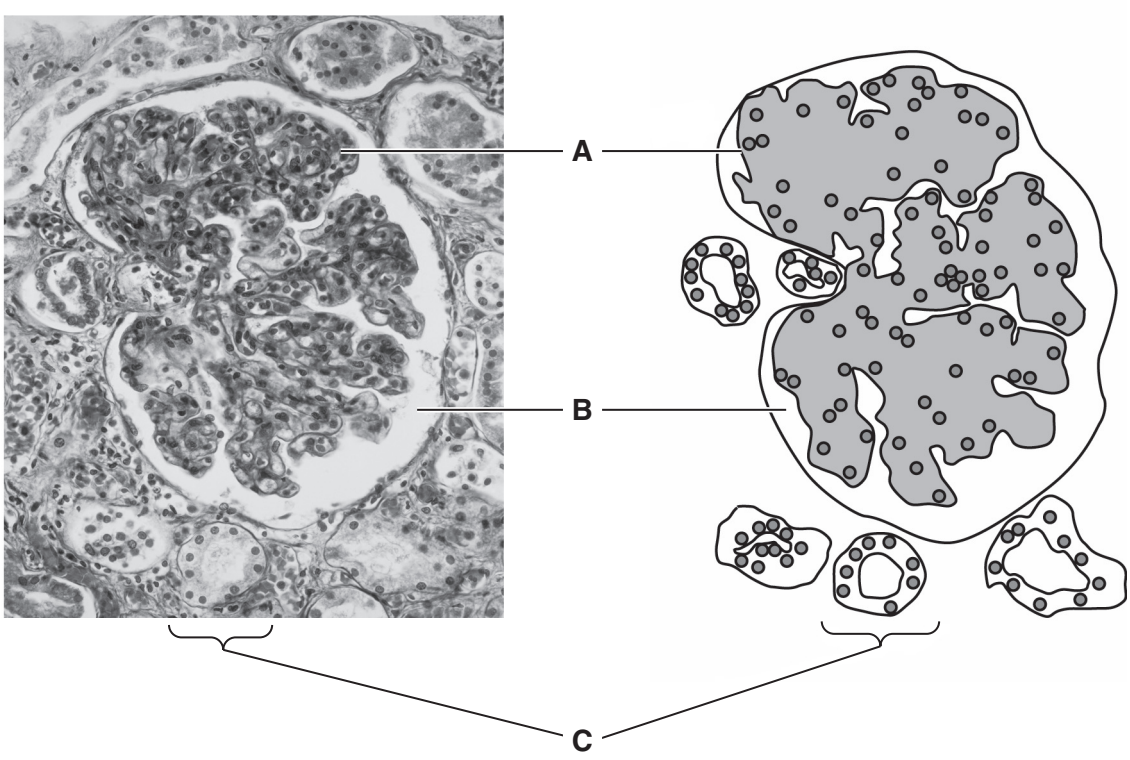

Fig. 1.1

(a) Identify the structures labelled **A** to **C** in Fig. 1.1.

A .....

B .....

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

(b) Nitrogenous waste is removed from blood plasma by the process of ultrafiltration.

Describe how the process of ultrafiltration takes place in the kidney **and** describe how structures in the kidney are adapted to allow ultrafiltration to occur.

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(c) One method of assessing kidney function is to determine the glomerular filtration rate (GFR). The GFR is the volume of fluid that is filtered out of the blood per minute.

Inulin is a polysaccharide that is used to determine the GFR.

- Inulin is removed completely from blood plasma by ultrafiltration.
- Inulin is **not** reabsorbed.
- Inulin is **not** secreted into the kidney tubules.

Inulin is given intravenously. The blood plasma concentration of inulin is then measured and urine samples are collected. The GFR can be determined from the concentration of inulin in a known volume of urine.

An inulin molecule is composed of a chain of 32 monosaccharide units. Fig. 1.2 shows the structure of inulin.

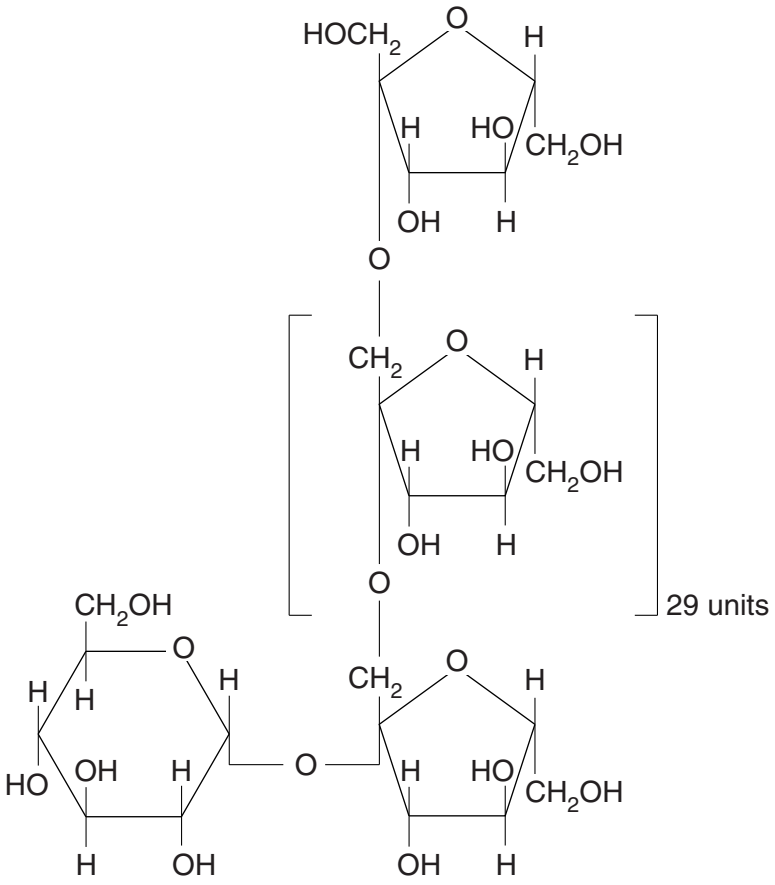

Fig. 1.2

(i) Name the bond joining the monosaccharide units in a molecule of inulin.

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

(ii) Suggest **two** properties of the inulin molecule that make it suitable for use in determining the GFR.

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

(d) Using inulin to determine the GFR is expensive. As an alternative, the GFR can be **estimated** from the blood plasma concentration of a substance called **creatinine**.

Creatinine is a breakdown product of creatine phosphate, a substance found in muscle cells.

Fig. 1.3 shows the relationship between plasma creatinine concentration and the GFR.

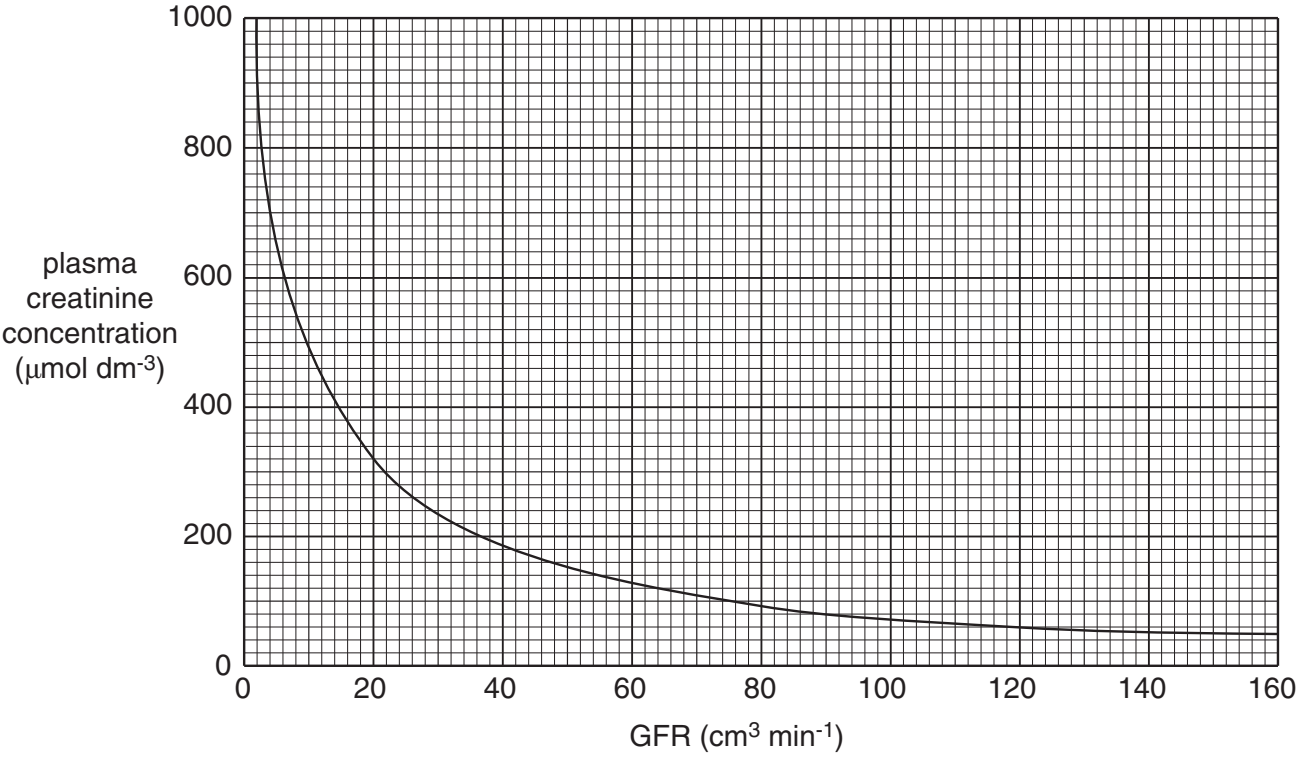

Fig. 1.3

(i) In normal kidney function, the GFR range is between 90 and 150 $cm^3\ min^{-1}$.

Estimate the normal range of **plasma creatinine concentration** using Fig. 1.3.

Range = ... **[2]**

(ii) Suggest why measuring the plasma creatinine concentration can only give an **estimate** of the GFR.

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

(e) When kidney function is severely impaired, renal dialysis may be necessary.

Outline the advantages and disadvantages of **haemodialysis** in the treatment of kidney disease.

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[Total: 21]

2 The ABO blood grouping in humans is determined by a single gene with three alleles: $\mathbf{I}^{A}$, $\mathbf{I}^{B}$ and $\mathbf{I}^{O}$.

(a) Distinguish between the terms *gene* and *allele*.

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QUESTION 2(b) STARTS ON PAGE 8

Fig. 2.1 shows a pedigree diagram of the inheritance of the ABO blood group. The blood group of some of the individuals is given in the pedigree.

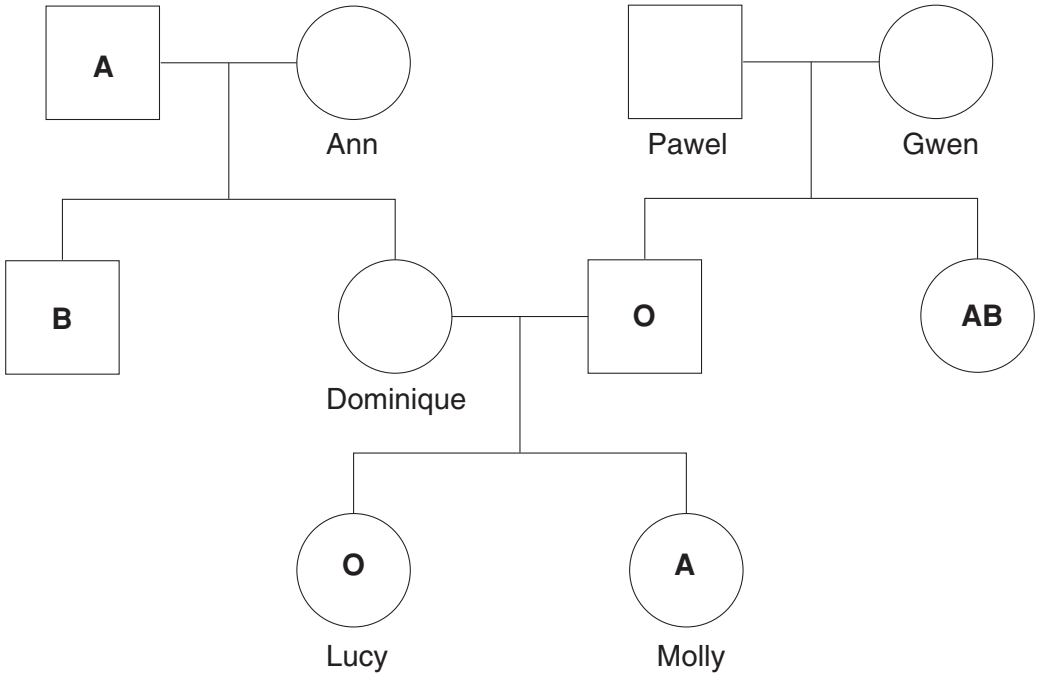

Fig. 2.1

(b) Using the information in Fig. 2.1 and the genetic symbols $\mathbf{I}^{A}$, $\mathbf{I}^{B}$ and $\mathbf{I}^{O}$, give the **phenotype** and **genotype** of the following individuals in the pedigree:

individual	phenotype	genotype
Dominique		
Ann		
Pawel		
Gwen		

[4]

(c) Fig. 2.1 shows that two sisters, Lucy and Molly, have different blood groups.

Explain why it is possible for Lucy's blood to be given to Molly, but Molly's blood cannot be given to Lucy.

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(d) In the past, the blood groups of parents and their offspring were compared as part of the evidence in disputes regarding the paternity of a child.

This technology has been replaced by **genetic profiling** (genetic fingerprinting).

- Genetic profiling requires the use of restriction enzymes.
- Restriction enzymes cut DNA to produce restriction fragments.
- Restriction fragments are labelled using probes that detect **minisatellites**.

(i) The restriction enzyme *BamH1* cuts DNA into fragments. *BamH1* cuts the DNA at the recognition site GGATCC.

Using *BamH1* as an example, explain what is meant by the term *palindromic sequence*.

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

(ii) Minisatellites are regions of DNA where a short sequence of bases is repeated in the same strand of DNA. The number of repeats varies from individual to individual.

Fig. 2.2 shows three DNA sequences, each with a different number of repeats.

- Each sequence contains two recognition sites for the restriction enzyme *BamH1*.
- *BamH1* cuts the DNA between the first and second G in the recognition site.

1	GGATCCCGATGATGATGATGATGATGATGATGATGGATCCCAATC
2	AAAGGCGGATCCCGATGATGATGATGAATCCGGTGGATCCCTTTTATT
3	AAATTACCCTTCGGATCCCGATGATGATGATGATGATGATGATGGATCC

Fig. 2.2

The length of a restriction fragment can be expressed as the number of **bases** contained in the fragment.

State the length of the largest restriction fragment that would be found in each DNA sample following treatment with *BamH1*.

1 …… bases

2 …… bases

3 …… bases **[3]**

(iii) Following the treatment of DNA with restriction enzymes, the restriction fragments are then analysed using gel electrophoresis.

A banding pattern is obtained that depends on the size of the restriction fragments.

Fig. 2.3 shows the banding pattern obtained from four blood samples in a paternity case.

The blood samples were taken from the child and three adults, **P**, **Q** and **R**. The three adults are the mother and two possible fathers.

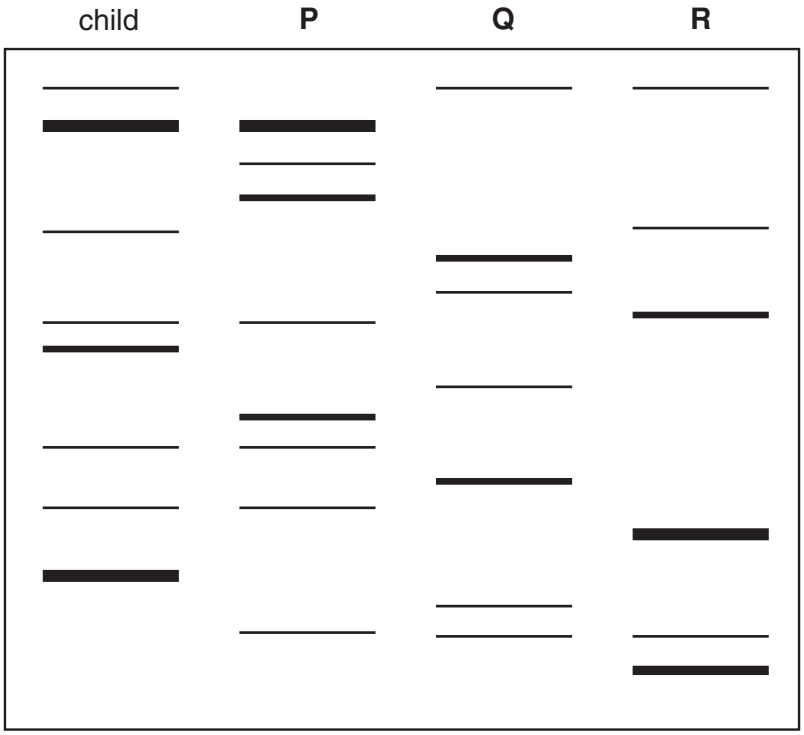

Fig. 2.3

Using the information in Fig. 2.3, state which of the letters **P**, **Q** and **R** identify the parents of the child.

Give reasons for your answer.

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

[Total: 19]

3 The following statement was issued by UK Transplant in September 2008.

> Between 1 April 2007 and 31 March 2008, 3235 organ transplants were carried out thanks to the generosity of 1665 donors.

(a) (i) Explain why the number of organ donations is greater than the number of organ donors.

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

(ii) Organs that are donated come from a wide range of sources.

Discuss the advantages and disadvantages of different sources of donated organs.

In your answer you should discuss a range of sources and give a critical consideration of each named source.

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QUESTION 3(b) STARTS ON PAGE 14

Cornea transplants account for approximately 40% of transplant operations each year.

During a cornea transplant operation, the damaged area of the existing cornea is removed and a section of healthy cornea from a donor is grafted into place.

(b) The cells that make up the cornea are kept transparent by the activity of a layer of endothelial cells. The endothelial cells form a layer between the cornea and the fluid in the eye, and actively remove water from the corneal cells.

Fig. 3.1 shows the relative positions of the cornea and the layer of endothelial cells.

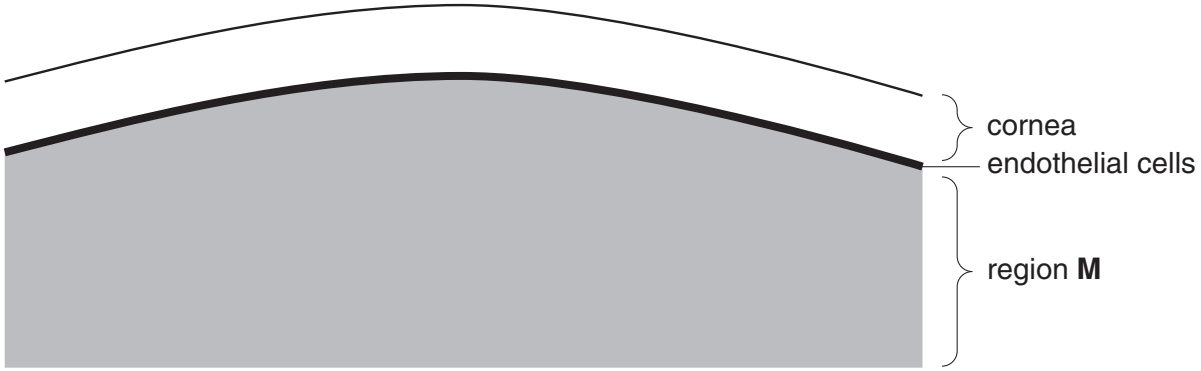

Fig. 3.1

(i) Name the fluid present in region **M**.

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

(ii) Explain why water moves into the corneal cells.

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

(c) There are no blood vessels in the cornea. Tissue matching is **not** essential for successful cornea transplants.

Explain why the lack of blood vessels in the cornea means that tissue matching is not essential for cornea transplants.

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

(d) Damage to the cornea may lead to a loss of visual acuity.

Fig. 3.2 shows a Snellen chart that is used to assess visual acuity.

Fig. 3.2

Suggest **two** variables that must be controlled during a visual acuity test in order to achieve valid results.

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

[Total: 17]

4 Thyroxine is a hormone secreted by the thyroid gland. Thyroxine has many important functions in the human body. One function is the control of metabolic rate.

(a) Fig. 4.1 shows how the secretion of thyroxine is controlled.

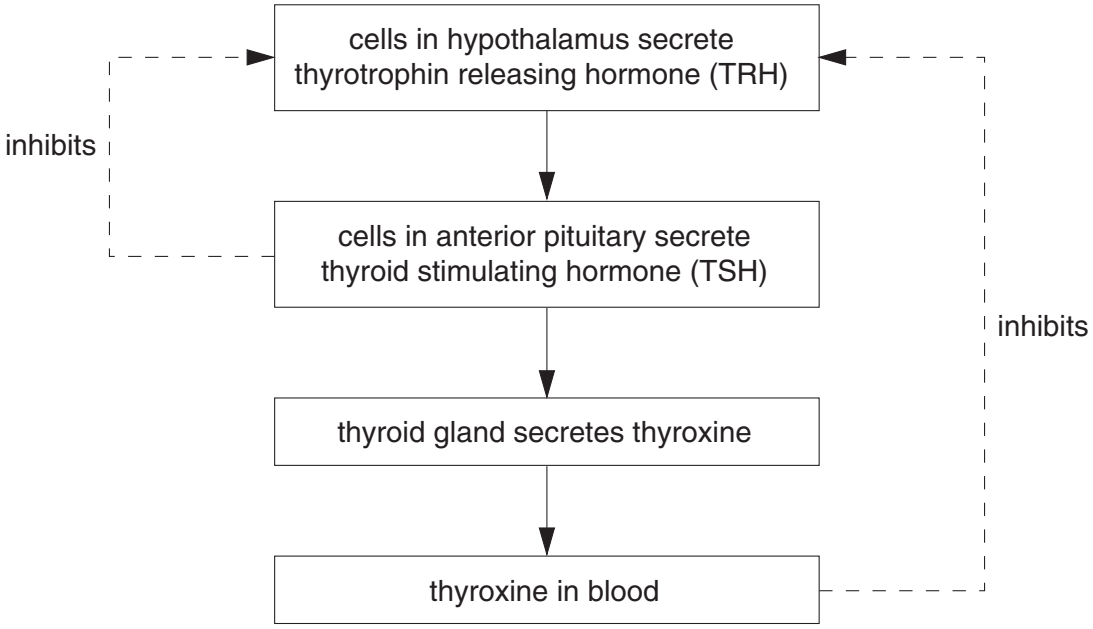

Fig. 4.1

Using the information in Fig. 4.1, explain how excessively high blood concentrations of thyroxine are prevented by negative feedback.

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

(b) One effect of an increase in blood thyroxine concentration is to increase the numbers of mitochondria present in some cells.

State **and** explain the long-term effect on the body of increasing the number of mitochondria present in cells.

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

QUESTION 4(c) STARTS ON PAGE 18

(c) One method of diagnosing an underactive thyroid involves measuring the concentration of TSH in blood samples and comparing these to the normal range.

TSH can be detected in a blood sample using an enzyme-linked immunosorbent assay (ELISA).

The steps in carrying out an ELISA are illustrated in Fig. 4.2.

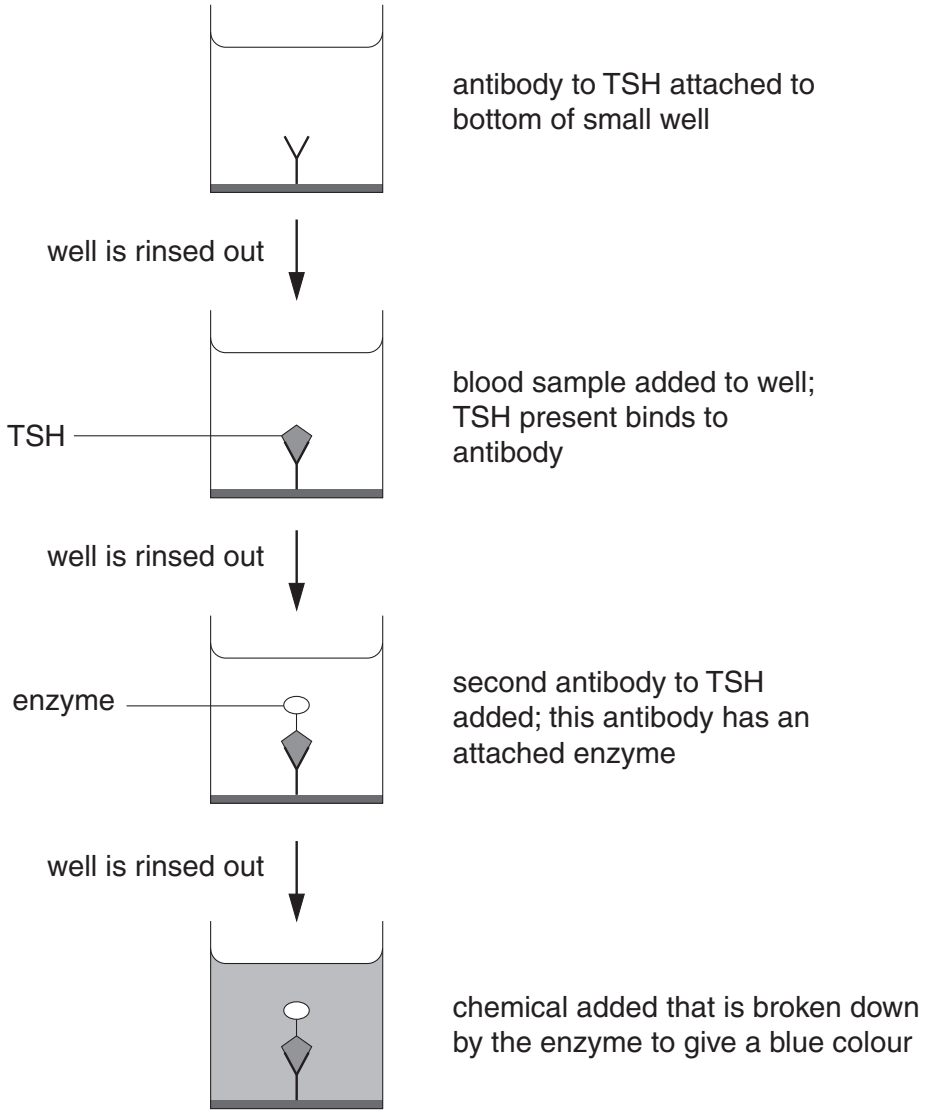

Fig. 4.2

The breakdown of the chemical by the enzyme produces a change in colour, which indicates the presence of TSH in the sample. Differences in colour intensity can be used to assess the concentration of TSH in the sample.

(i) Complete the following passage that describes in detail how the ELISA works.

TSH consists of two protein chains that are modified in the …… of the cells of the anterior pituitary to form a glycoprotein. The TSH in the blood sample acts as an …… and binds to the …… region of the antibody. The second antibody binds to the TSH. The chemical added in the final stage of the procedure acts as the …… for the enzyme attached to the second antibody. This chemical binds to the …… of the enzyme to form an …… . **[6]**

(ii) Suggest why it is important to rinse out the well between each stage of the procedure.

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

[Total: 14]

5 Osteoporosis can be diagnosed by the use of a bone density scan (bone densitometry).

(a) Outline how a bone density scan is carried out.

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(b) Osteoporosis can cause small fractures of vertebral bones resulting in a loss of height and a curved back.

Table 5.1 shows the prevalence of vertebral fractures associated with osteoporosis in males and females of different ages in the United Kingdom.

Table 5.1

age (years)	prevalence of vertebral fractures (cases per 10 000 persons)		female prevalence ÷ male prevalence × 100
	males	females	
50 – 54	1 350	699	52
55 – 59	1 500	938	63
60 – 64	1 660	1 230	74
65 – 69	1 810	1 570	87
70 – 74	1 980	1 980	100
75 – 79	2 140	2 460	115
80 – 84	2 300	3 000	130
85+	2 630		165

(i) Using the information in Table 5.1, calculate the prevalence of vertebral fractures in women aged 85 or over.

Show your working and give your answer to the nearest whole number.

Answer = ... cases per 10 000 persons **[2]**

(ii) Compare and contrast the prevalence of vertebral fractures in males and females.

In your answer you should use data to support statements.

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(iii) Suggest **one** way in which osteoporosis treatment in the 50 – 59 age group would differ between males and females. Give a reason for your answer.

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(c) As part of routine monitoring of bone health, blood samples are taken and the concentration of calcium is measured.

Calcium is present in blood as the calcium ion (Ca^{2+}) and bound to the plasma protein albumin. Low blood concentration of calcium is a risk factor for osteoporosis.

(i) Outline the **dietary** advice that should be offered to people to maintain their blood calcium concentration.

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(ii) Suggest **one** further consequence of low blood calcium concentration **other than** the effect on bone health. Give a reason for your suggestion.

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[Total: 16]

6 **(a)** The presence of the myelin sheath speeds up the transmission of the nerve impulse in myelinated neurones.

Outline how a nerve impulse is transmitted along a myelinated neurone.

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The lower jaw in humans contains the lingual nerve and the inferior dental nerve.

When teeth in the lower jaw require dental treatment, an anaesthetic is injected. The injection is located so that the lingual nerve and the inferior dental nerve are both affected. This causes numbness in the jaw and also in parts of the tongue and cheek. The tongue can still be moved.

(b) Suggest what type of neurones in the lingual and inferior dental nerves are affected by the anaesthetic. Give a reason for your suggestion.

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

QUESTION 6(c) STARTS ON PAGE 24

(c) The anaesthetic molecule diffuses into the neurone. The molecule then combines with hydrogen ions in the cytoplasm. This enables the molecule to bind to the sodium ion channels which prevents the passage of sodium ions.

Suggest why the anaesthetic molecule was only able to bind to the sodium ion channel **after** it had combined with hydrogen ions.

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(d) Following dental treatment, painkilling drugs such as opioids may be given.

Outline how opioids block the transmission of pain in the central nervous system.

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

[Total: 13]

END OF QUESTION PAPER