

Friday 15 June 2012 – Morning

A2 GCE HUMAN BIOLOGY

F225 Genetics, Control and Ageing

Candidates answer on the Question Paper.

OCR supplied materials:

- Insert (inserted)

Other materials required:

- Electronic calculator
- Ruler (cm/mm)

Duration: 2 hours



Candidate forename		Candidate surname	
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Centre number						Candidate number				
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INSTRUCTIONS TO CANDIDATES

- The Insert will be found in the centre of this document.
- 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 **28** pages. Any blank pages are indicated.

Answer **all** the questions.

1 The central nervous system consists of the brain and spinal cord.

Fig. 1.1 is a diagram of a spinal reflex arc.

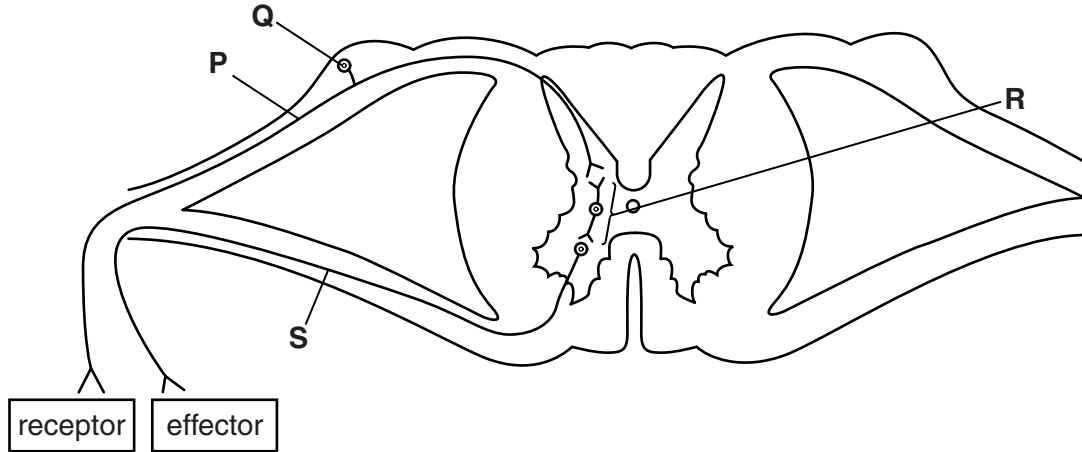


Fig. 1.1

(a) Identify the structures labelled **P** to **S** in Fig. 1.1.

- P**
- Q**
- R**
- S** [4]

(b) Testing reflexes is one way of assessing damage to the central nervous system, including damage caused by traumatic brain injury.

(i) State what is meant by the term '*traumatic brain injury*'.

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(ii) Describe how the **results** of an MRI scan can be used to assess traumatic brain injury.

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[Total: 10]

2 Sickle cell anaemia is an inherited disease affecting haemoglobin.

Fig. 2.1, **on the insert**, shows a photomicrograph of a blood smear taken from a person with sickle cell anaemia.

(a) Using the information in Fig. 2.1, outline the **evidence** that supports the following statements:

(i) the blood smear has come from a person with sickle cell anaemia

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(ii) the blood smear was stained using a differential stain.

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(b) Gel electrophoresis can be used to separate pieces of DNA on the basis of their molecular size.

Small DNA fragments travel further along the electrophoresis gel than larger fragments.

- The restriction enzyme *Mst II* recognises the sequence CCTNAGG – where N can be any DNA nucleotide.
- Sickle cell anaemia is caused by a mutation in the β -globin gene.
- The mutation changes CCTG**A**GG to CCTG**T**GG.
- Mutated DNA produces **longer** fragments of DNA.

(i) Using *Mst II* as an example, explain what is meant by a restriction enzyme.

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(ii) Explain why longer fragments of DNA occur in the mutated form of the β -globin gene.

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(c) Samples of DNA from three individuals were treated with the restriction enzyme *Mst II* and then loaded onto an electrophoresis gel.

- Individual **F** is homozygous for normal haemoglobin and has the normal β -globin gene.
- Individual **G** is homozygous for sickle cell anaemia and has the mutated form of the β -globin gene.
- Individual **H** is heterozygous.

Following electrophoresis, the DNA was located using a radioactive probe.

Fig. 2.2 shows a diagram of the electrophoresis gel and the relative positions of the DNA fragments for individuals **F** and **G**.

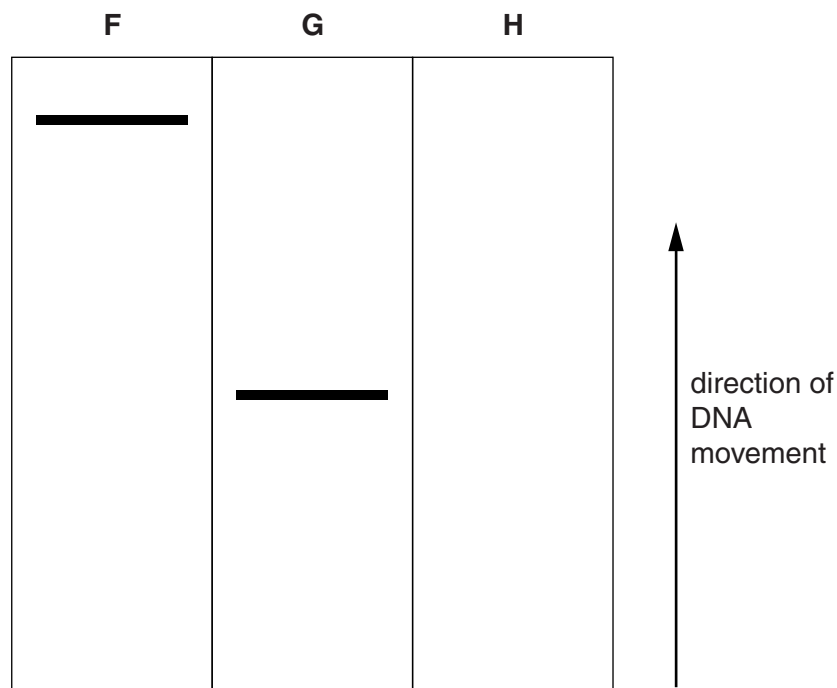


Fig. 2.2

Complete Fig. 2.2 by **drawing the position of the DNA** from individual **H** in the blank column on the diagram.

..... *The answer to this question must be written on Fig. 2.2* [1]

(d) State the term used to describe the pattern of inheritance shown by sickle cell anaemia and normal haemoglobin alleles.

..... [1]

- (e) Complete the following passage which compares the structure and properties of fetal and adult haemoglobin.

Fetal haemoglobin is a globular protein consisting of four polypeptide chains. It is said to have a structure. When the dissociation curves for adult and fetal haemoglobin are compared, the curve for fetal haemoglobin is positioned to the left of that for adult haemoglobin. This is because fetal haemoglobin has a higher for than adult haemoglobin. [3]

- (f) In some Indian populations, people who are homozygous for the sickle cell allele show much milder forms of sickle cell anaemia.

- An analysis of blood from these populations shows that 25% of the haemoglobin present is fetal haemoglobin.
- Fetal haemoglobin does **not** become crystallised at low partial pressures of oxygen.
- Fetal haemoglobin differs in structure to adult haemoglobin. Fetal haemoglobin consists of two alpha (α) and two gamma (γ) polypeptide chains.
- Some chemicals such as hydroxyurea (**HU**) can switch on the gamma haemoglobin gene in adult bone marrow stem cells.
- Double-blind randomised trials have been carried out to assess the effectiveness of HU in treating sickle cell anaemia.

- (i) With reference to trials to evaluate a treatment for sickle cell anaemia, explain what is meant by the terms 'double-blind' and 'randomised'.

double-blind.....
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randomised.....
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..... [2]

Table 2.1 summarises the results from one double-blind randomised trial, carried out over two years, using **HU** to treat sickle cell anaemia.

Table 2.1

factor being measured	result for individuals given HU	result for individuals given placebo
fetal haemoglobin at beginning of trial (%)	5.0	5.2
fetal haemoglobin after 2 years (%)	8.6	4.7
admissions to hospital for sickle cell crisis treatment over 2 years (%)	2.4	3.9
number of blood transfusions over 2 years	55	79
number of deaths related to sickle cell anaemia	2	4
number of neutrophils ($\times 10^9 \text{ dm}^{-3}$) after 2 years	4.9	6.4

- (ii) It was calculated that the individuals who were given **HU** treatment showed a 72% increase in fetal haemoglobin over the two year period of the trial.

Using the information in Table 2.1, show how a value of 72% was calculated.

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(iii) Using the information in Table 2.1, describe the evidence that supports the hypothesis that increasing the percentage of fetal haemoglobin in an individual reduces the symptoms of sickle cell anaemia.

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(iv) Suggest **why** the number of neutrophils is higher in individuals with sickle cell anaemia who were given the placebo rather than in those who were given **HU**.

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[Total: 23]

3 The light sensitive region of the eye is the retina.

Fig. 3.1, **on the insert**, shows a computer generated picture of cells in the human retina.

The cell labelled **A** represents a rod cell.

(a) (i) Using the letters, **B, C, D** or **E**, identify the following cells in Fig. 3.1.

name of cell	letter
ganglion cell	
bipolar cell	

[2]

(b) Colour vision is a function of the cone cells in the retina. Colour blindness can occur due to genetic mutations in the genes for the pigments in cone cells.

- Red-green colour blindness is a sex linked recessive condition inherited in the same way as haemophilia.
- Yellow-blue colour blindness is an autosomal recessive condition inherited in the same way as cystic fibrosis.

(i) State what is meant by the term ‘sex linkage’.

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..... [2]

(ii) The following statements may apply to red-green colour blindness **or** to yellow-blue colour blindness **or** to both.

Complete the following table by inserting either the letter **R**, **Y** or **B** for each of the statements.

- R** – red-green colour blindness
- Y** – yellow-blue colour blindness
- B** – both types of colour blindness

statement	letter
colour blind fathers can pass on the condition to their sons	
a female who is not colour blind could be homozygous or heterozygous	
a heterozygous normal female and a male with colour blindness who have a child have a 25% chance of having a colour blind son	
males are more likely to be colour blind than females	

[4]

(iii) Children are routinely tested for colour blindness.

Outline how a test for colour blindness is carried out **and** suggest why it is important to diagnose colour blindness at an early age.

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(c) Changes in the eye due to ageing can also affect colour vision. Two examples of changes which can lead to a loss or reduction in colour vision are macular degeneration and cataracts.

Suggest why macular degeneration and cataracts can lead to loss or reduction of colour vision.

macular degeneration.....

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cataracts.....

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..... [3]

[Total: 22]

4 Many people are concerned about the level of alcohol consumption in the United Kingdom.

The following statements describe some of the effects of alcohol on the body.

- J** Some people find that drinking alcohol is more important than anything else they might do during the day.
- K** Alcohol is a depressant but, as it acts on the limbic system in the brain, the effect is to reduce inhibitions and anxieties.
- L** One serious alcohol withdrawal symptom is called delirium tremens. This includes shaking, sweating, diarrhoea and seizures and requires urgent medical attention as it can be life threatening.
- M** Delerium tremens cause you to feel agitated, confused, paranoid and experience hallucinations.

(a) With reference to alcohol consumption, explain what you understand by the term 'drug' and explain the difference between physical and psychological dependency.



In your answer you should make use of the statements to support each of your explanations.

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QUESTION 4(b) STARTS ON PAGE 16

(b) Breakdown of alcohol in liver cells reduces the amount of pyruvate available for conversion to glucose by gluconeogenesis. This reduces the amount of glucose produced by the liver to maintain glucose levels.

(i) State **two** ways by which pyruvate can be produced in liver cells.

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(ii) Suggest why Type 1 diabetics are advised not to drink alcohol unless they eat at the same time.

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17
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PLEASE DO NOT WRITE ON THIS PAGE
QUESTION 4(c) STARTS ON PAGE 18

(c) Fig. 4.1(a) shows how **mortality** due to alcohol related events changes with age.

Fig. 4.1(b) shows how the **causes** of alcohol related deaths change with age over the same time period.

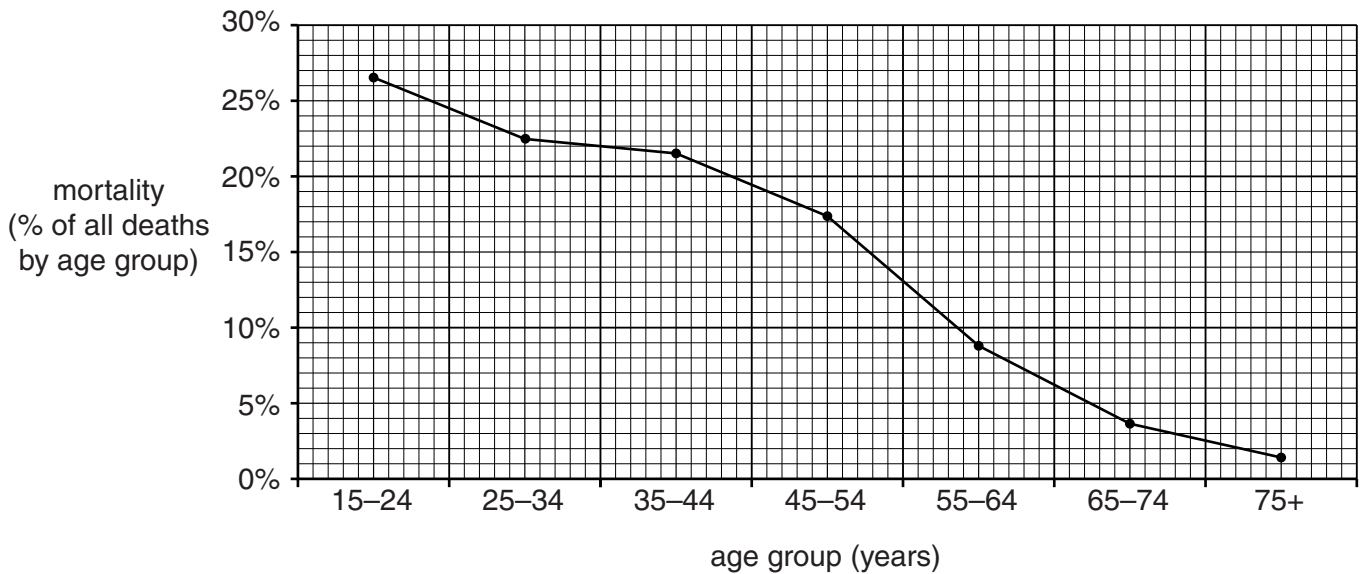
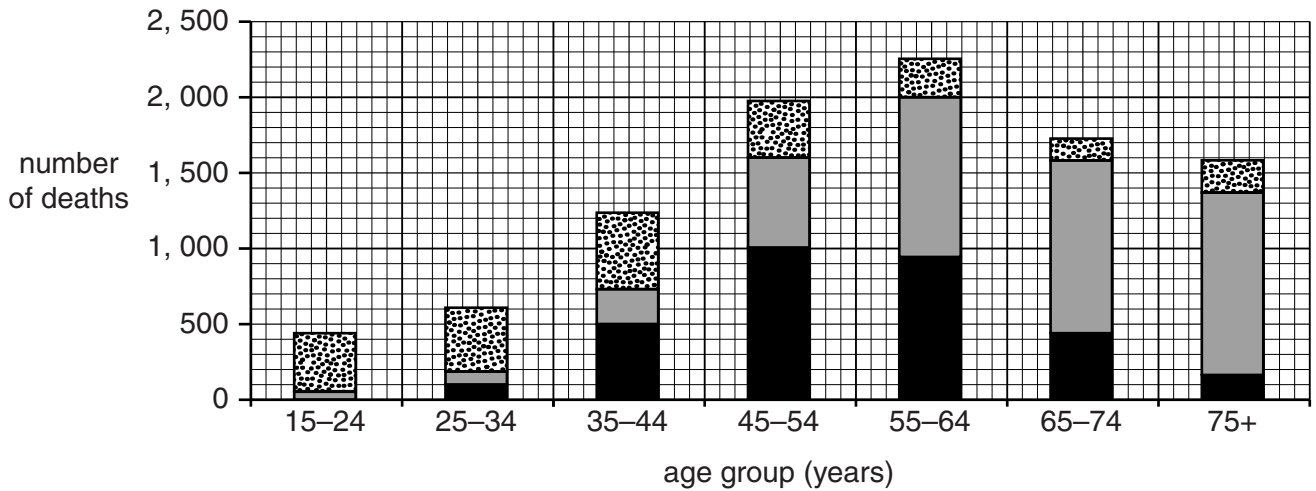


Fig. 4.1(a)



Key:

- alcohol related deaths due to acute events e.g. road traffic accident
- alcohol related deaths due to chronic events e.g. liver disease
- deaths caused directly by alcohol

Fig. 4.1(b)

- (i) Using the information in Fig. 4.1(a) and Fig. 4.1(b), suggest why the percentage of deaths due to alcohol related events, such as road traffic accidents, declines with age even though the number of deaths due to alcohol increases.

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- (ii) Using Fig. 4.1(b), compare the pattern of alcohol related deaths in the age groups 15–24 and 55–64 years.

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[Total: 17]

5 Blood pressure is regulated in several ways.

One mechanism involves renin (angiotensinogenase).

The role of renin in regulating blood pressure is outlined in Fig. 5.1.

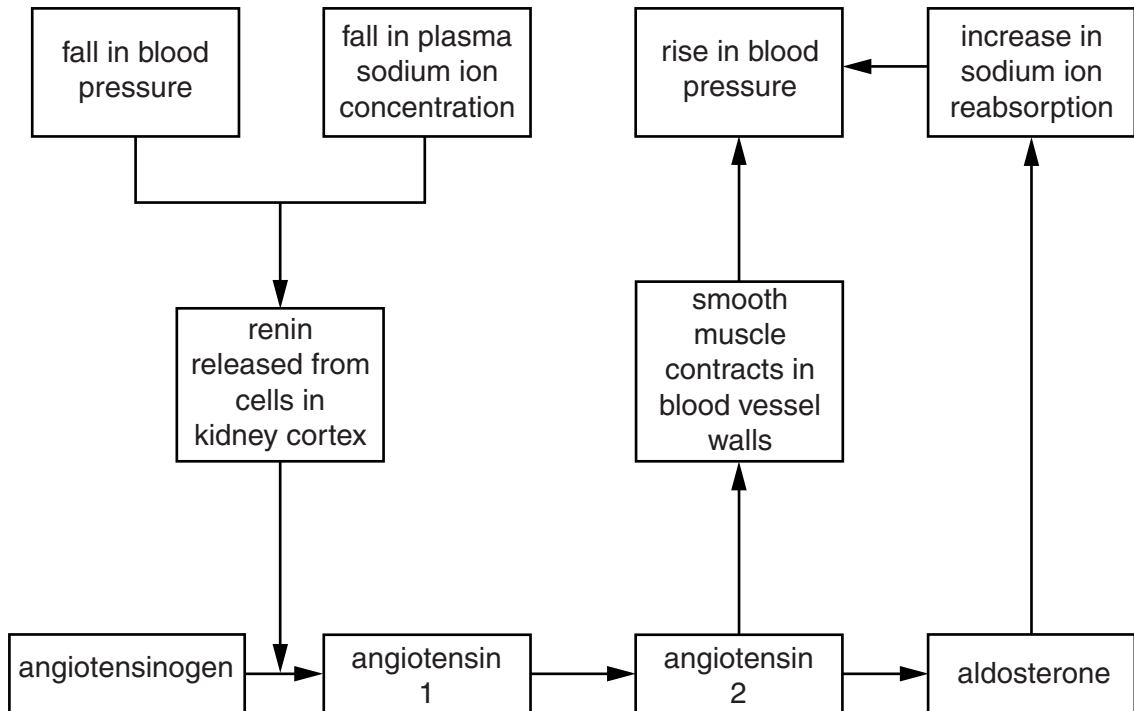


Fig. 5.1

(a) Using the information in Fig. 5.1, explain why control of blood pressure is an example of homeostasis.

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(b) Aldosterone is a **steroid** hormone which is produced by the adrenal cortex. Release of aldosterone stimulates an increase in the reabsorption of sodium ions in the kidney.

(i) Suggest the **precise** location of the receptors for aldosterone in the kidney.

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(ii) Explain why an increase in the reabsorption of sodium ions causes an increase in blood pressure.

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QUESTION 5(c) STARTS ON PAGE 22

(c) Angiotensinogen is a polypeptide consisting of 452 amino acids that is produced by liver cells.

- Renin acts on angiotensinogen to produce angiotensin 1.
- Angiotensin 1 consists of a chain of 10 amino acids.

A student wrote a short passage describing the action of renin on angiotensinogen.

'The bonds between the amino acids in angiotensinogen are broken by a condensation reaction. It happens because renin is an enzyme (angiotensinogase). Angiotensinogen has a shape which is identical to the active site on the enzyme. It binds to renin to form an enzyme substrate complex and the ester bonds between the amino acids are broken. This results in the formation of the substrate angiotensin 1.'

Unfortunately, four of the terms used by the student in the passage were used incorrectly.

In the table below, identify the terms which were used incorrectly and insert a more appropriate term in the box provided. The first one has been done for you.

incorrect term	appropriate term
condensation	hydrolysis

[6]

(d) Fig. 5.1, on page 20, shows that angiotensin 1 is converted to angiotensin 2. Angiotensin 2 binds to calcium ion channels on smooth muscle cells causing the cells to contract.

Suggest how the binding of angiotensin 2 **initiates** smooth muscle contraction.

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..... [2]

[Total: 18]

QUESTION 6 STARTS ON PAGE 24

6 Changes due to ageing can occur in several different body systems, including the reproductive system.

Births in Japan were monitored over a period of one year. During this period, the age of the parents at the time of the child's birth was recorded.

The results of this study are shown in Fig. 6.1.

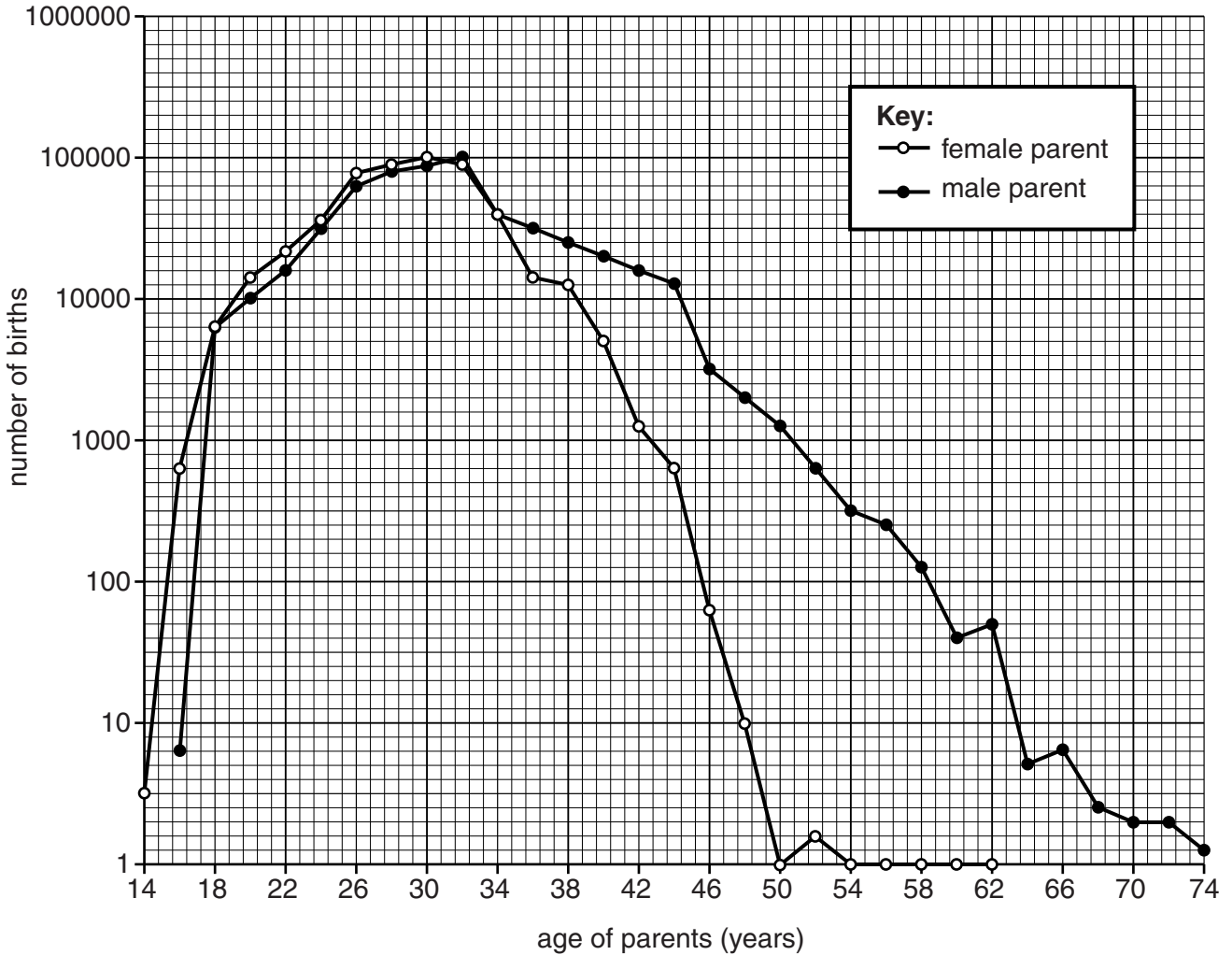


Fig. 6.1

(a) Explain the difference in the pattern of data for male and female parents between the ages of 35 and 50 years.

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(b) Suggest **two** reasons why no births were recorded for fathers under the age of 16.

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(c) The cardiovascular and respiratory systems also show changes with increasing age.

(i) On average, the cardiac output at rest of a 35 year old man is $5.8 \text{ dm}^3 \text{ min}^{-1}$.

On average, a 75 year old man at rest has 30% less cardiac output than a 35 year old man.

Calculate the cardiac output of an average 75 year old man.

Give your answer to one decimal place.

Answer = [2]

(ii) State **three** changes that occur in the respiratory system that are due to ageing.

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[Total: 10]

END OF QUESTION PAPER

ADDITIONAL PAGE

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

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