

Monday 21 January 2013 – Morning

A2 GCE HUMAN BIOLOGY

F225/01 Genetics, Control and Ageing

Candidates answer on the Question Paper.

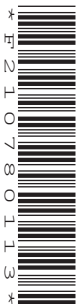
OCR supplied materials:

- 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 page 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 **24** pages. Any blank pages are indicated.

Answer **all** the questions.

1 Human erythrocytes (red blood cells) carry many different antigens on their cell surface membranes. One class of these antigens determines a person's ABO blood group. There are four ABO blood group phenotypes.

(a) (i) Explain what is meant by the term *phenotype*.

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..... [1]

(ii) Using **one** of the ABO blood groups as an example, explain how two different **genotypes** can result in the same phenotype.

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- (b) The gene which determines the ABO blood group of a person is located on chromosome 9. The gene responsible for nail patella syndrome is also located on this chromosome.

Fig. 1.1 is a diagram showing an event which takes place during meiosis.

The letters **D** to **H** represent the loci of some of the genes present on chromosome 9.

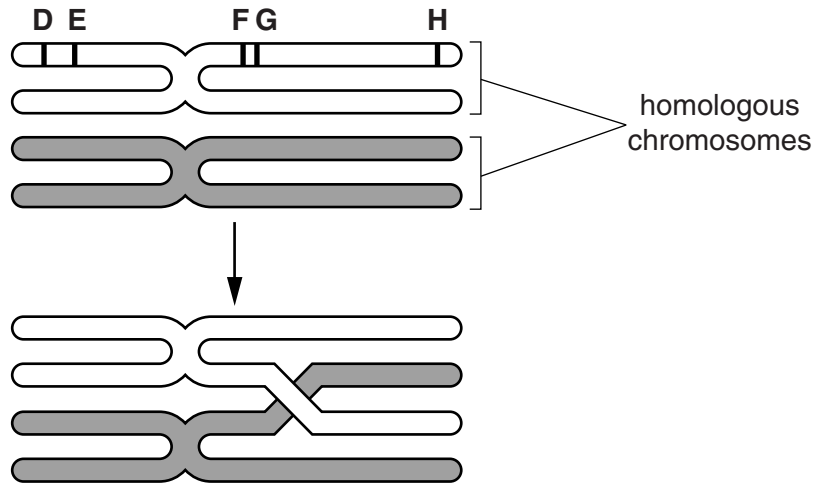


Fig. 1.1

- (i) State precisely when in meiosis the event shown in Fig. 1.1 would take place.
 [2]
- (ii) Letter **G** represents the locus of the gene controlling the ABO blood groups.
 Which letter is most likely to identify the locus of the gene responsible for nail patella syndrome?
 letter [1]
- (iii) Using the information in Fig. 1.1, explain what is meant by the term *autosomal linkage*.

 [3]

- (c) The gene responsible for nail patella syndrome codes for a 'transcription factor'.

A transcription factor binds to a region of DNA close to the beginning of a gene, allowing the gene to be 'switched on'.

Name two **types** of molecule that will be produced by the cell after the transcription factor binds to the DNA.

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..... [2]

- (d) In addition to abnormalities with nail and patella development, one further feature associated with nail patella syndrome is a form of glaucoma.

Describe the **symptoms** of glaucoma and the changes in the eye that cause these symptoms.

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..... [3]

- (e) Individuals with nail patella syndrome can also show symptoms of abnormal development and functioning of the basement membrane in the glomerulus. These symptoms are detected by analysis of urine samples.

Suggest and **explain** how the abnormal development and functioning of the basement membrane might affect the composition of urine.

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..... [3]

[Total: 19]

5
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QUESTION 2 STARTS ON PAGE 6

- (b) The United Kingdom's Foresight Programme is operated by the Government Office for Science. The aim of the programme is to model future trends so that effective strategies can be developed and implemented.

One area that has been investigated is obesity. Some of the data used in the model to assess the impact of future trends in obesity in males aged between 21 and 60 years is shown in Table 2.1 and Fig. 2.1, **on the insert**.

Table 2.1, below, shows the relative risk of developing type 2 diabetes for a given BMI group. The data are for males aged between 21 and 60 years.

Table 2.1

BMI	Relative risk of developing type 2 diabetes
0–23	1.0
23–24	1.0
24–25	1.5
25–27	2.2
27–29	12.0
29–31	30.0
31–33	40.0
33–35	55.0
35+	90.0

Fig. 2.1, **on the insert**, shows how the proportion of the male population (aged between 21 and 60 years) in each BMI group is predicted to change.

In Fig. 2.1, the 'zone' on either side of the plotted lines represents the 'confidence limits' of the data.

- (i) In the period between 1993 and 2003, which BMI categories show the most **reliable** data?

Give a reason for your suggestion.

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..... [2]

- (ii) Describe what happens to the **confidence limits** between 2003 and 2050 for data in the BMI category 25–30.

Suggest a reason for the pattern you have described.

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- (iii) What evidence is there in the data in Fig. 2.1 and Table 2.1 to support the prediction that type 2 diabetes will become increasingly common?

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- (iv) Identify **two** problems with the presentation of the independent variable in Table 2.1 and its use in Fig. 2.1.

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..... [2]

- (c) Certain types of food, when eaten regularly, can increase the risk of developing type 2 diabetes. To reduce this risk, health professionals recommend eating foods with a low glycaemic index (**GI**).

The **GI** of a food is a measure of how much the blood glucose concentration is increased after the food has been eaten. Glucose has a value of 100 and the **GI** value of other foods is given as a percentage of this.

Another useful measure is the glycaemic load (**GL**) of a food. This takes into account both the **GI** and the portion size (mass) of the food eaten. The **GL** can be calculated using the formula given below.

$$\text{GL} = \frac{\text{GI}}{100} \times \text{mass of digestible carbohydrate per serving}$$

- (i) Use the formula for **GL** to complete **Table 2.2** below.
Give each answer to the nearest whole number.

Table 2.2

Food type	GI (glycaemic index)	Mass of carbohydrate per serving (g)	Mass of digestible carbohydrate (g)	GL (glycaemic load)
Basmati rice	58	150	38
Brown rice	66	150	21

[2]

- (ii) Foods with a low glycaemic load (**GL**) may also reduce the risk of developing cancer of the colon.

Using the information in Table 2.2, suggest why foods with a low **GL** may reduce the risk of developing cancer.

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..... [2]

[Total: 23]

- 3 The pupil controls the amount of light which enters the eye. The size of the pupil can be adjusted in response to changes in light intensity or as a result of emotions such as excitement or fear.
- (a) Complete the following passage concerning the structure of the eye, and the nerves controlling the size of the pupil.

The pupil is a circular hole in a sheet of muscle known as the

Light entering the pupil passes through the lens and then through the before striking an area of the retina called the This area contains the greatest concentration of light receptor cells known as cells.

The muscle fibres around the pupil are arranged as circular muscles and radial muscles. Both of these are involuntary muscles and are controlled by the nervous system. In dim light or in response to excitement, the radial muscles contract in response to nerve impulses from a motor neurone and the pupil dilates. In bright light, the circular muscles contract in response to nerve impulses from a motor neurone so the pupil constricts.

The pupil response is rapid and automatic and is known as a

[8]

(b) If the brain and optic nerves are functioning correctly, a light shone into **one** eye should produce the same pupil constriction in **both** eyes.

To assess the pupil response it is recommended that

- the lighting in the room is dim
- the light source is shone first on one eye, and then on the other eye. This is repeated several times.

(i) Suggest a reason for these recommendations when carrying out the pupil response test.

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..... [2]

(ii) Fig. 3.1(a) shows pupils at the beginning of a pupil response test.

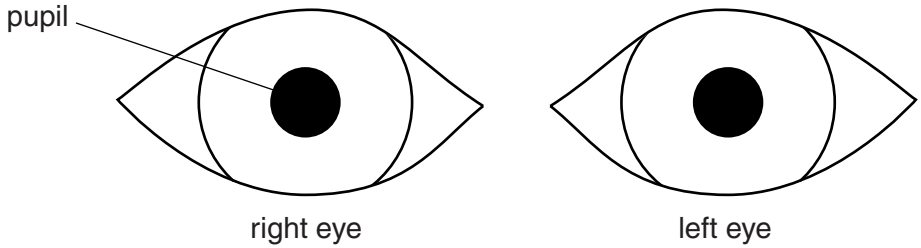


Fig. 3.1(a)

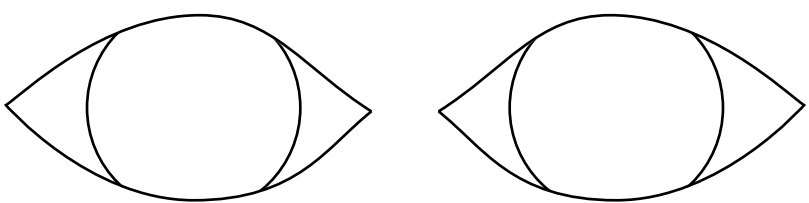


Fig. 3.1(b)

Draw on Fig. 3.1(b) above, the appearance of **both pupils** if a light is shone on the left eye when there is a **normal** pupil response. [1]

(c) The diagrams in Fig. 3.2 below show an **abnormal** pupil response observed in a patient.

Fig. 3.2(a) shows the appearance of both pupils when light is shone in the right eye.

Fig. 3.2(b) shows the appearance of both pupils when light is shone in the left eye.

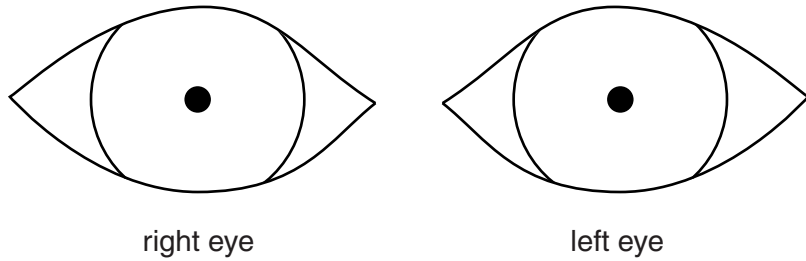


Fig. 3.2(a)

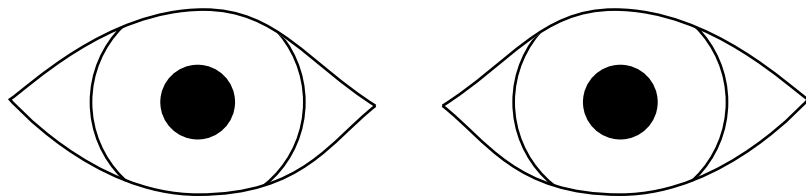


Fig. 3.2(b)

The consultant ophthalmologist concluded that, rather than having brain damage, the patient had a detached retina in their left eye. A detached retina occurs when the retina is no longer in contact with the underlying layer in the eye.

Suggest why a detached retina may lead to the pupil reflex no longer happening.

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..... [2]

(d) Observing the pupil response is one way in which a person’s level of consciousness can be assessed. State one further test, **other than the pupil response test**, that could be used to assess the level of consciousness.

..... [1]

[Total: 14]

Turn over

4 One way of treating a patient with kidney failure is to use haemodialysis.

Fig. 4.1 is a simplified diagram of a haemodialysis machine.

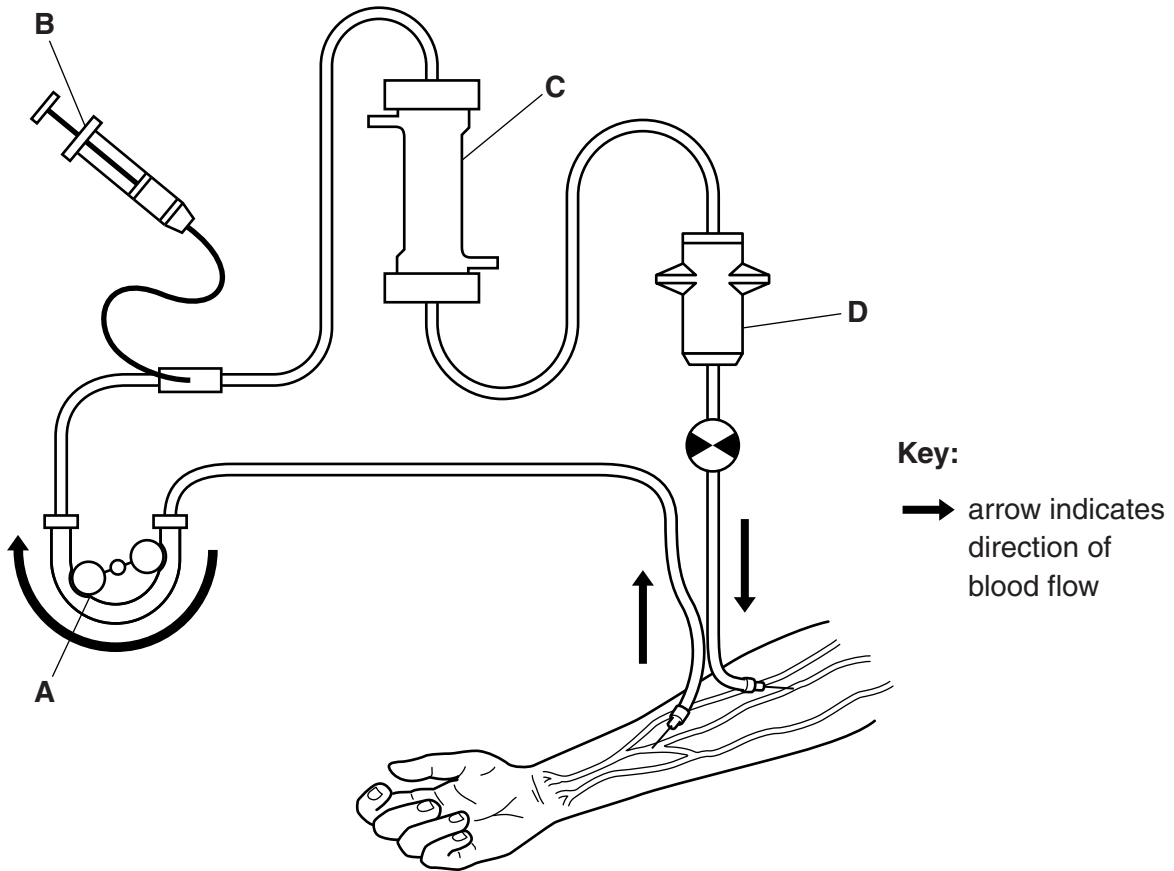



Fig. 4.1

(a) Describe and explain how the haemodialysis machine is used in the treatment of kidney failure.

 In your answer you should make clear the roles of the structures labelled **A**, **B**, **C** and **D** in Fig. 4.1.

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(b) Patients on haemodialysis are advised to limit their intake of foods that are high in phosphate.

- Phosphate ions can combine with calcium ions to form mineral deposits which can build up in blood vessel walls.
- High levels of phosphate in the diet can lead to high blood pressure.

Suggest why high levels of phosphate in the diet can lead to high blood pressure.

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..... [2]

(c) Clinical trials have been carried out in which sodium citrate is added to blood while a patient is undergoing haemodialysis.

Suggest **one** advantage of adding sodium citrate to the blood of a patient on haemodialysis. Explain the reason for your suggestion.

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..... [2]

[Total: 14]

(b) Outline **two** possible **social** consequences of hearing loss for an elderly person.

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..... [2]

(c) When the results from an audiology test show significantly more hearing loss in one ear than the other, one possible explanation is a tumour.

One type of tumour which produces this effect develops on the balance nerve and is called a **Schwannoma**.

Suggest the **precise** location of a Schwannoma tumour in a neurone **and** outline how the presence of such a tumour could be confirmed.

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..... [3]

[Total: 9]

6 Endorphins are important in cell signalling and are thought to act by binding to receptors on neurones.

- Endorphins stimulate the release of a neurotransmitter called dopamine from neurones in the brain.
- Dopamine is responsible for feelings of happiness and wellbeing.

(a) Explain how neurotransmitters carry out their role in the nervous system.

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..... [3]

(b) Diamorphine (heroin) is known to bind to the endorphin receptors on neurones.

(i) Suggest why diamorphine can bind to endorphin receptors.

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..... [1]

(ii) Suggest why a person taking heroin may develop a psychological dependency.

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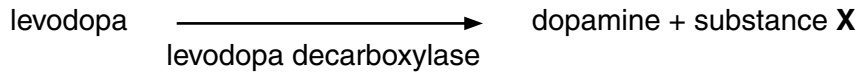
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..... [2]

- (c) Dopamine has an important role as a neurotransmitter in an area of the brain which coordinates movement.

If the neurones which normally synthesise dopamine fail to function, the drug **levodopa** can be given. Levodopa can be taken up by neurones and converted to dopamine.

The word equation below summarises how levodopa is converted into dopamine.



- (i) State what **type** of protein levodopa decarboxylase is and suggest what substance **X** is.

type of protein

substance **X** [2]

- (ii) State the name **and** outline the symptoms of a condition which levodopa could be used to treat.

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..... [3]

(d) Endorphins are small polypeptide molecules consisting of short chains of amino acids.

Sequencing the human genome has led to the identification of the sequences of some of the genes coding for small polypeptides.

(i) Explain why a gene coding for a small polypeptide of 9 amino acids would be **predicted** to contain 27 nucleotides.

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..... [2]

(ii) It was found that the actual length of the gene which codes for the endorphin molecule was considerably longer than 27 nucleotides.

Suggest **two** reasons why the gene coding for the endorphin molecule is longer than 27 nucleotides.

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..... [2]

[Total: 15]

- 7 In January 2011, a transplant of embryonic stem cells was carried out on Nina, a child born with a rare, life threatening genetic condition. The source of the stem cells was her brother.

Fig. 7.1 summarises the stages which occurred before the stem cell transplant.

Step 1	Nina’s parents discussed the options available for treatment with health professionals
Step 2	Nina’s parents underwent IVF treatment resulting in the production of several embryos
Step 3	embryos were screened
Step 4	a suitable embryo was identified and implanted into Nina’s mother
Step 5	the baby, a boy, was born and embryonic stem cells were harvested from his umbilical cord
Step 6	embryonic stem cells were transplanted from the baby to his sister Nina.

Fig. 7.1

- (a) In **Step 1**, one of the health professionals involved in the discussions was a genetic counsellor.

Outline the role of the genetic counsellor in the discussions with Nina’s parents.

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..... [2]

- (b) In addition to the embryos being screened for genetic diseases, suggest **one further** genetically controlled feature which may have been screened for in **Step 3**. Explain the reason for your suggestion.

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..... [2]

(c) The embryonic stem cells used in this case were **not** obtained by therapeutic cloning.

Suggest what is meant by 'therapeutic cloning'.

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..... [2]

[Total: 6]

END OF QUESTION PAPER

ADDITIONAL ANSWER SPACE

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

A large rectangular area with horizontal dotted lines for writing, intended for providing additional answers. The area is bounded by a solid line on the left and bottom, and dotted lines on the top and right.



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