

Tuesday 11 June 2013 – 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	
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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 **28** pages. Any blank pages are indicated.

Answer **all** the questions.

1 (a) HRT is used to treat symptoms of the menopause.

Combined HRT involves taking both oestrogen and progesterone.

Describe the difference between combined HRT and **continuous** combined HRT.

.....

.....

.....

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.....

.....

[3]

(b) Many investigations have been carried out into the effect of HRT on women's health. Some investigations such as the 'Million Women' study have been observational studies. Other investigations have involved the use of clinical trials.

Fig. 1.1 shows the results of one investigation into the effects of HRT on a range of possible conditions.

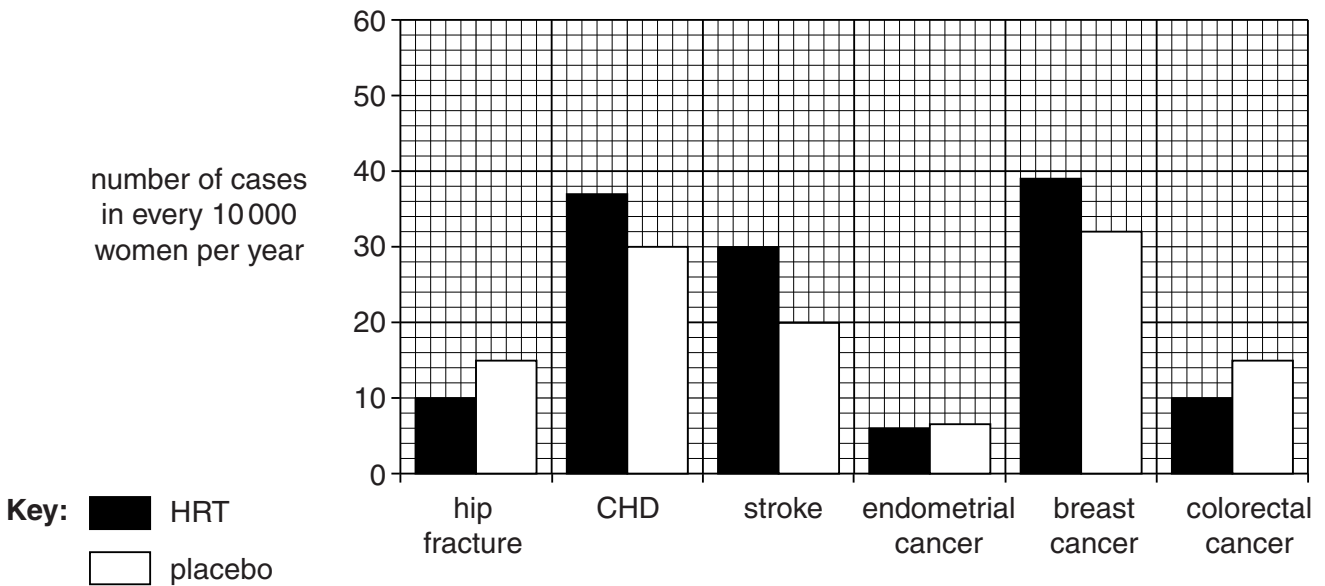


Fig. 1.1

State **one** piece of evidence from Fig. 1.1 which indicates that these results come from a clinical trial rather than an observational study.

.....

.....

[1]

(d) Suggest why the following women were not included in the sample for this investigation:

- women who had undergone a hysterectomy

.....

.....

.....

.....

.....

- women whose blood pressure was routinely measured at or above 140/90mmHg (18.6/12kPa).

.....

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.....

.....

[3]

[Total: 12]

5
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- 2 Nerve impulses occur due to changes in both the concentration and location of sodium and potassium ions inside and outside neurones.

Fig. 2.1 represents a cross section of an axon at rest in a **myelinated** neurone.

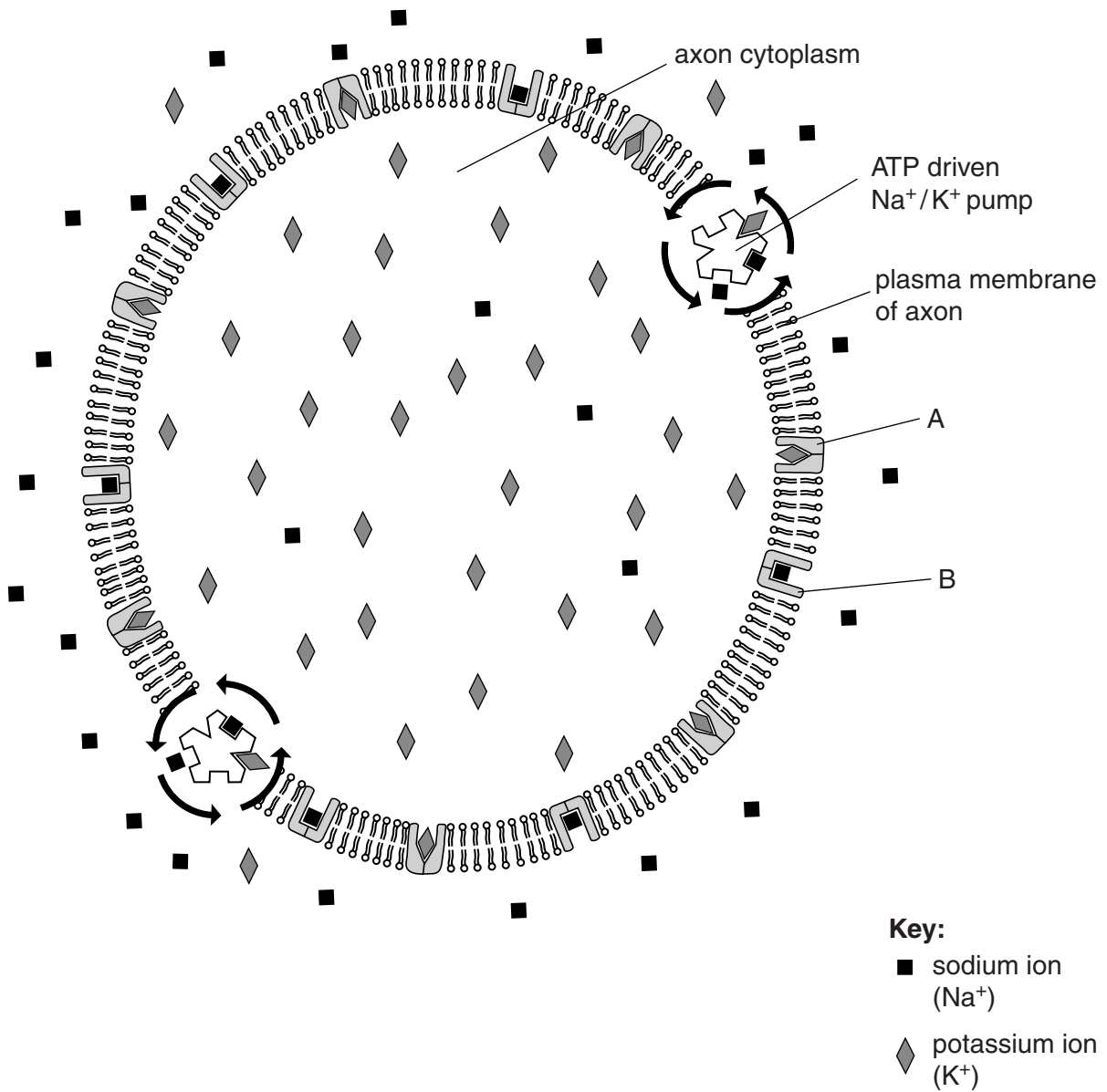


Fig. 2.1

(a) (i) State the main component of the plasma membrane of the axon shown in Fig. 2.1.

..... [1]

(ii) Using Fig. 2.1, identify **one** piece of evidence that indicates that the cross section through the axon is at a node of Ranvier.

.....

 [1]

(b) Table 2.1 below summarises the position of two types of channels, A and B (open or closed), the movement of ions and the activity of the ATP driven Na⁺/K⁺ pump **during an action potential**.

Complete the table to show what is happening during **depolarisation** and **repolarisation** of the axon.

	At rest	Depolarisation	Repolarisation
Channel A position	closed		
Channel B position	closed		closed
Sodium ion diffusion	no diffusion	rapid diffusion in	
Potassium ion diffusion	some diffusion out		
ATP driven Na⁺/K⁺ pump	active		active

Table 2.1

The answers to question 2(b) should be written in Table 2.1.

[5]

Question 2(c) begins on page 8

(c) Repolarisation of an axon results in a refractory period.

- During the refractory period, the axon is in a condition known as *hyperpolarisation*.
- During this time no further depolarisation is possible at the region of the axon which is hyperpolarised.
- No further action potentials can occur in a hyperpolarised region.

(i) Using the information in Fig. 2.1 and Table 2.1, suggest why no depolarisation is possible during the refractory period.

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

(ii) Outline the significance of the refractory period in the transmission of the nerve impulse in a myelinated neurone.

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

(d) Refractory periods are also seen in muscle fibres. During the refractory period in muscle fibres, no new contraction can be initiated.

- In skeletal muscle, the refractory period is between 1 and 2 milliseconds.
- In cardiac muscle, the refractory period is 200 milliseconds.

Suggest why a relatively long refractory period is necessary for cardiac muscle to carry out its role.

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

(e) Axons which are damaged in the peripheral nervous system may regenerate and transmit nerve impulses again. However, axons which are damaged in the central nervous system usually fail to regenerate resulting in loss of communication between neurones.

(i) Identify a test which could be carried out to establish that peripheral nerves have completely regenerated.

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

(ii) Outline **two** reasons why neurones in the central nervous system fail to regenerate.

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

[Total: 16]

3 Haemophilia A is a genetic disease caused by a mutation in the gene coding for a clotting factor known as Factor VIII. Factor VIII is a glycoprotein which is absent in the plasma of people with haemophilia A.

(a) (i) State the location in a human cell of the Factor VIII gene.

.....
 [2]

(ii) Give **two** reasons why the Golgi apparatus is essential for the release of Factor VIII into blood plasma.

.....

 [2]

(b) Fig. 3.1 is a pedigree diagram showing how haemophilia is inherited.

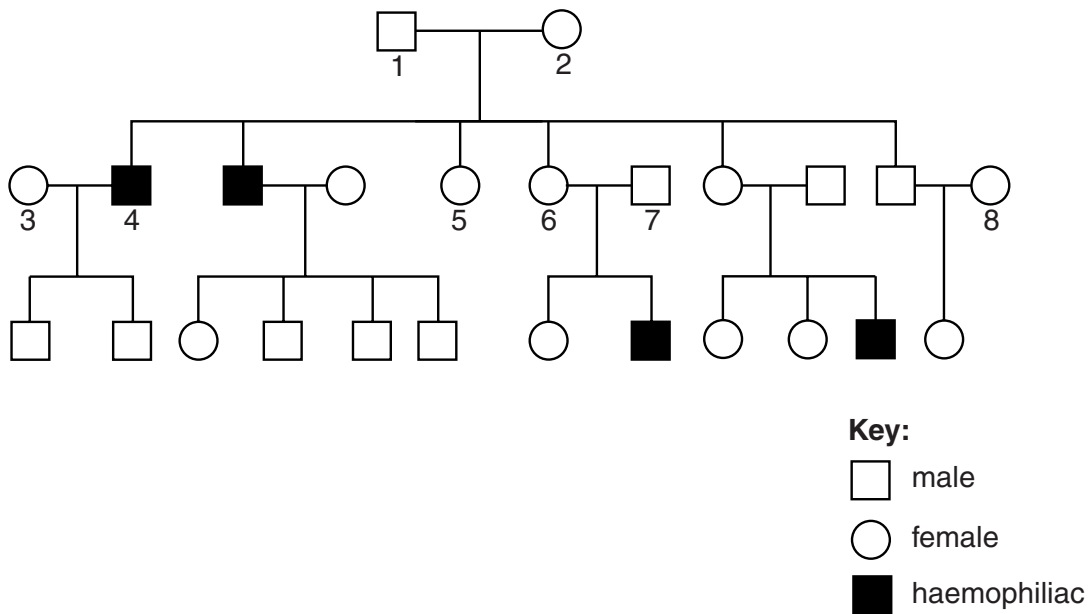


Fig. 3.1

- (i) Complete the table below which matches up an individual from the pedigree with their corresponding genotype. The first one has been completed for you.

Number of the individual	Genotype
3 and 8	$X^H X^H$
4	
6	
7	

[3]

- (ii) What is the probability of individual 5 having the same genotype as individuals 3 and 8?

Explain your reasoning.

probability =

explanation

.....

.....

..... [3]

Question 3(c) begins on page 12

(c) Maternally inherited diabetes and deafness (MIDD) is a rare form of diabetes.

- MIDD is caused by a mutation in a gene in the **mitochondrial DNA**.
- MIDD tends to develop between the ages of 25 and 30.
- Treatment of MIDD requires insulin injections.
- People diagnosed with MIDD usually have a BMI of less than 30.

(i) Identify which of the features of MIDD are similar to type 1 diabetes and which are similar to type 2 diabetes.

type 1

.....

type 2

..... [3]

(ii) MIDD occurs in approximately equal numbers in males and females.

Suggest why MIDD can be inherited by both males and females but is only passed on by females.

.....

.....

.....

.....

..... [2]

[Total: 15]

13
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- 4 Corneal transplants are sight-saving operations with a high success rate. In the year 2008, a total of 2711 people in the United Kingdom had their sight restored through the NHS Blood and Transplant service.

The most common reason for corneal transplants in younger people is a condition called **keratoconus**. In this condition, the shape of the cornea changes.

Fig. 4.1(a) shows a normal cornea and Fig. 4.1(b) shows a cornea with keratoconus.

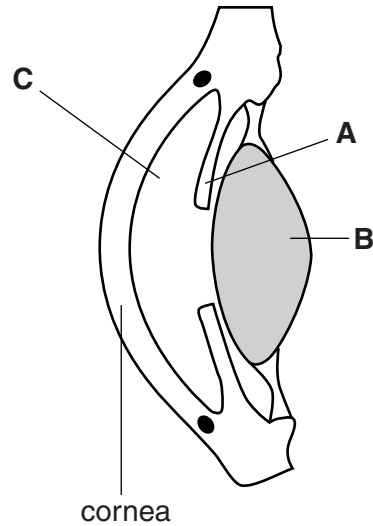


Fig. 4.1(a)

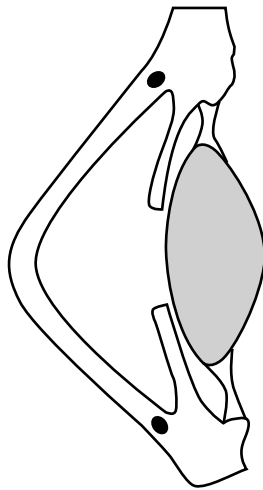


Fig. 4.1(b)

(a) (i) Identify the structures labelled **A**, **B** and **C** on Fig. 4.1(a).

A

B

C

[3]

(ii) Explain why vision is affected if the shape of the cornea is changed.

.....

.....

.....

.....

..... [2]

(b) Corneal transplants were given to 2711 patients in 2008. After one year, the transplants were still functioning in 2512 of these patients.

Calculate the percentage of transplant patients for whom the operation was **not** successful after one year.

Show your working. Give your answer to one decimal place.

Answer =% [2]

[Total: 7]

(c) The kidneys are important organs in homeostasis. The presence of erythrocytes (red blood cells) in urine is **one** indication of kidney (renal) failure.

(i) State two further **symptoms** of kidney failure.

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

(ii) State two possible **causes** of kidney failure.

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

(iii) A person with kidney failure may require a kidney transplant.

State **two** possible sources of a donated kidney.

1
2 [2]

(d) Erythrocytes rarely appear in the urine of a person with kidney failure who also has diabetes insipidus.

Suggest why.

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

[Total: 20]

- 6 DNA profiling relies on the existence of ‘minisatellites’. These are short sequences of DNA, usually between 2 and 5 base pairs long, that repeat many times. They are also referred to as short, tandem repeats or **STRs**.

One STR that is used in genetic profiling occurs on chromosome 7. The STR base sequence is GATA and the number of repeats can vary from 6 to 15.

Fig. 6.1 is a diagram showing the DNA of an individual who is **heterozygous** for this STR. Each small square represents one repeat.

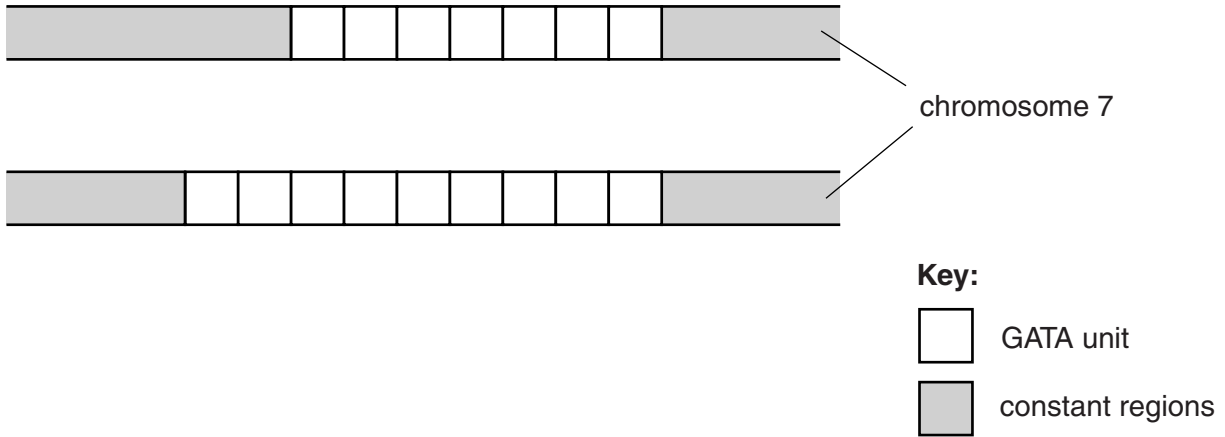


Fig. 6.1

- (a) Write out the sequence of bases on the DNA strand that is complementary to one GATA unit.

Answer = [1]

- (b) In the polymerase chain reaction (PCR), the enzyme DNA polymerase is used to make multiple copies of regions of DNA.

The reaction mixture includes the sample of DNA to be copied plus the following ingredients:

- DNA primers – short pieces of DNA which bind to the constant regions at both ends of the sequence to be copied
- buffer solution
- heat-stable DNA polymerase (Taq polymerase)
- deoxynucleotides (deoxyATP, deoxyTTP, deoxyCTP and deoxyGTP)

- (i) Suggest why a buffer needs to be present in the reaction mixture.

.....

 [2]

- (ii) The deoxynucleotides that are added to the reaction mixture are the monomers used for making the new DNA strands.

Suggest **one further** reason for adding the deoxynucleotides to the reaction mixture.

..... [1]

(c) In the first stage of PCR, the mixture is heated to a temperature of around 90°C.

At 90°C

- the DNA in the sample ‘denatures’ as the two strands separate
- the Taq polymerase does **not** denature.

(i) Suggest why high temperatures are needed to separate the two DNA strands.

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

(ii) Suggest why, unlike most enzymes, the Taq polymerase molecule is **not** denatured at 90°C.

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

(d) A student made the following prediction:

The temperature needed to denature the DNA would be higher than 90°C if the repeating sequence was **GCGC** instead of *GATA*.

(i) Suggest how the student justified this prediction.

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

(ii) Suggest one factor, **other than the composition of the reaction mixture**, that the student would need to control to carry out a valid experiment to test the prediction in (d).

Explain why this factor needs to be controlled.

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

(ii) A child has parents who are both heterozygous at the GATA locus on chromosome 7.

- One parent has 8 copies and 12 copies of the GATA repeat.
- The second parent has 15 copies and 8 copies of the GATA repeat.

The DNA from the child was used to produce a DNA profile (banding pattern).

How many of the possible banding patterns for the child will show as **two** bands?
You may find it helpful to use the grid provided below.

Answer = [2]

[Total: 22]

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7 The following passage is taken from the 2009 World Alzheimer Report.

Dementia is a syndrome due to disease of the brain, usually chronic, characterised by a progressive deterioration in intellect including memory, learning, orientation, language, comprehension and judgement.

- (a) Dementia syndrome is linked to a large number of underlying brain pathologies including Alzheimer's disease and vascular dementia.

Table 7.1 shows some of the changes associated with Alzheimer's disease and vascular dementia.

Type of dementia	Early characteristic symptoms	Changes in brain tissue	Proportion of dementia cases (%)
Alzheimer's disease (AD)	impaired memory, apathy and depression with gradual onset	build up of protein X outside neurones forming plaques and build up of protein Y inside neurones forming neurofibrillary tangles	50–75
Vascular dementia (VD)	similar to AD , but memory less affected, mood fluctuations more prominent, symptoms appear in sudden steps	cerebrovascular disease due to deterioration of the blood vessels supplying the brain	20–30

Table 7.1

- (i) With reference to the information on Alzheimer's disease in Table 7.1, identify the two proteins **X** and **Y**.

X

Y [2]

(ii) Suggest **two** risk factors which might increase the chances of developing **vascular dementia**.

1

.....

2

.....

[2]

(iii) Using the information in Table 7.1, suggest why dementia due to Alzheimer’s disease has a gradual onset whereas dementia due to vascular disease can develop in sudden steps.

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..... [2]

(b) State **two** ways in which damage to the brain tissue can be assessed by health professionals.

1

2

[2]

[Total: 8]

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 solid line on the left side, creating a margin. The rest of the page is filled with horizontal dotted lines, providing space for writing. The lines are evenly spaced and extend across the width of the page.

ADDITIONAL ANSWER SPACE

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