Centre Number

Candidate Number

cea
Rewarding Learning

ADVANCED
General Certificate of Education
2019

Life and Health Sciences

Assessment Unit A2 5
assessing
Genetics, Stem Cell Research and Cloning

MV18

[AZ051]
THURSDAY 6 JUNE, AFTERNOON

Time

1 hour 45 minutes, plus your additional time allowance.

Instructions to Candidates

Write your Centre Number and Candidate Number in the spaces provided at the top of this page.
Write your answers in the spaces provided in this question paper.
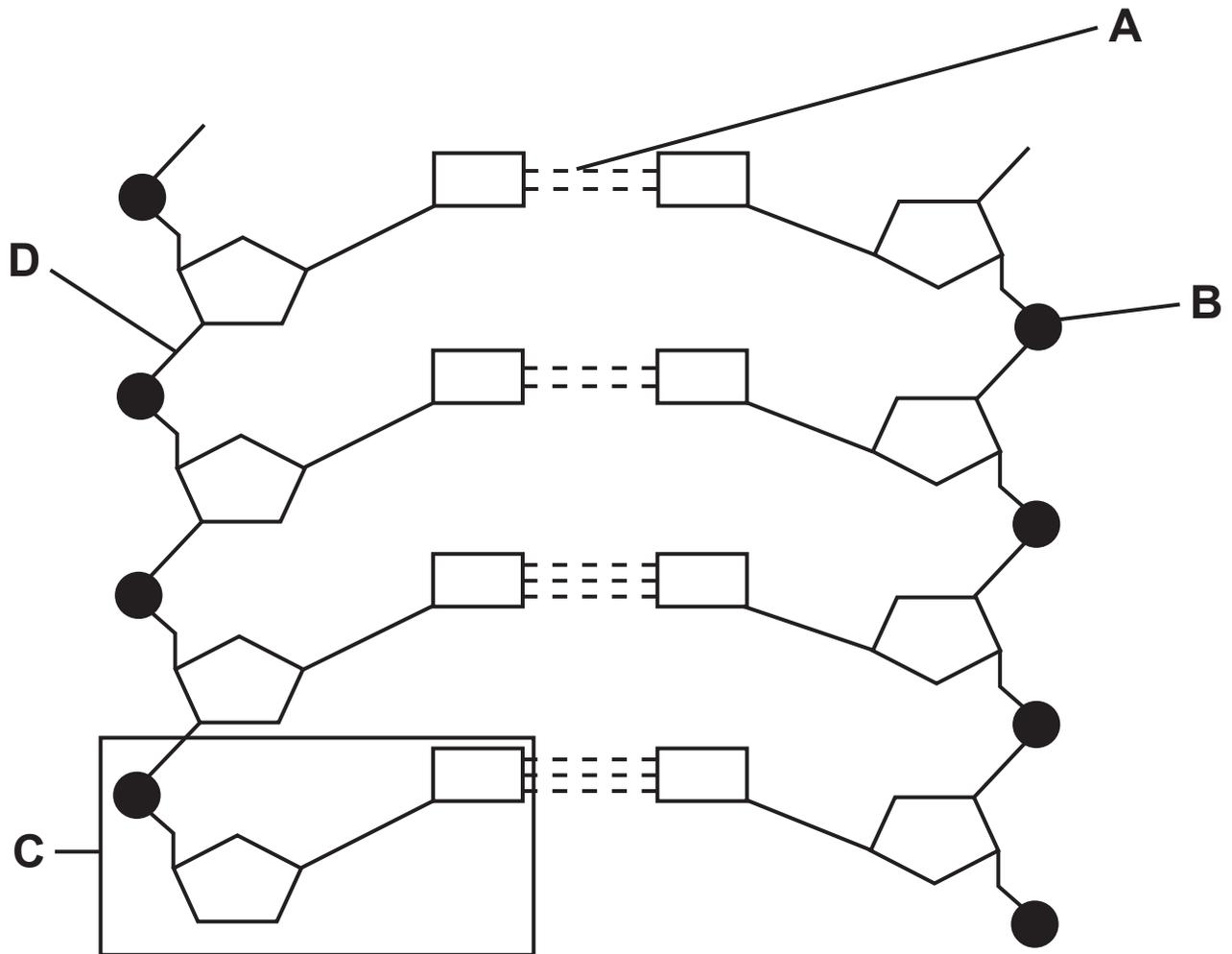Answer **all nine** questions.

Information for Candidates

The total mark for this paper is **100**.
Figures in brackets printed at the end of each question indicate the marks awarded to each question or part question.
Quality of written communication will be assessed in Question **5(b)**.

1 The diagram below represents the structure of DNA.

(a) Scientists Watson and Crick described the structure of DNA as a double helix.

   (i) Name **one** other scientist who added to our understanding of the structure of DNA.
   Describe the contribution of this scientist.
   [2 marks]

   Name _____

   Description of contribution _____

   _____

   _____

(ii) Name the parts of the diagram labelled **A**, **B**, **C** and **D**.
[4 marks]

A _____

B _____

C _____

D _____

In an analysis of the DNA in the nucleus of a cell, 23**%** of the bases were found to be guanine.

(iii) Calculate the percentage of each of the other bases in the DNA.   [3 marks]

You are advised to show your working.

Adenine _____ **%**

Cytosine _____ **%**

Thymine _____ **%**

Another nucleic acid found in cells is ribonucleic acid (RNA).

(iv) State **two differences** between the structure of DNA and RNA.   [2 marks]

1. _____

_____

_____

2. _____

_____

_____

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(Questions continue overleaf)

2 The steps **A** to **E** below describe how DNA replication occurs.

They are **not** in the correct order.

A Each strand now acts as a template.

B DNA polymerase joins adjacent nucleotides on the new strands.

C The enzyme DNA helicase unwinds the two strands of DNA.

D Free DNA nucleotides enter opposite their complementary bases.

E Hydrogen bonds between complementary bases are broken.

(a) Use the letters to put the steps in the correct order in the boxes below.   [3 marks]

The first step is given.

| C | → | | → | | → | | → | |

(b) Three theories existed to describe the process of DNA replication.

**(i)** Complete the table below by naming the three theories.   [1 mark for each]

Name of Theory	Description of DNA replication process
	In a new DNA molecule, one strand is conserved from the parent molecules while the other strand is newly synthesised.
	The parent molecule is unchanged. The complete parent DNA molecule acts as a template for the new daughter molecule.
	The parent molecule breaks into segments. New nucleotides fill in the gaps precisely.

(ii) Name the theory which is commonly accepted today and the two scientists who provided evidence to support this theory.   [2 marks]

Theory _____

Scientists _____ and _____

3 The diagram below shows stem cells and the cells that they can form.

Stem cells

Red blood cells

Cardiac muscle cells

Nerve cells
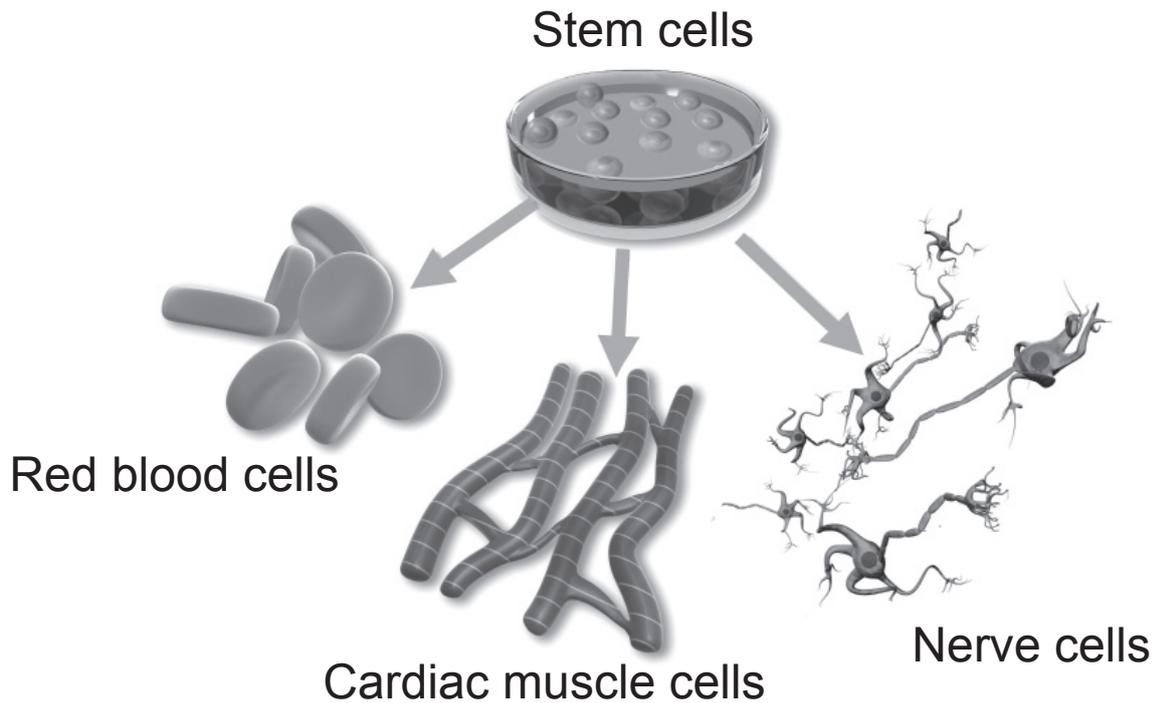
(a) (i) Using your knowledge and the diagram above, describe **two** key features of stem cells.   [2 marks]

1. _____

_____

2. _____

_____

(ii) State **two differences** between embryonic and adult stem cells.   [2 marks]

1. _____

_____

2. _____

_____

Scientists are investigating the use of embryonic stem cells in the treatment of patients with diabetes.

Patients with diabetes cannot regulate their blood glucose levels.

(b) (i) Suggest the type of cell the embryonic stem cell could become in a patient with diabetes.   [1 mark]

_____

(ii) Suggest how these cells are of benefit to a person with diabetes.   [1 mark]

_____

4 Cystic fibrosis is caused by a mutated allele of the gene for the production of the protein CFTR.

(a) (i) Describe and explain the effect of cystic fibrosis on the respiratory system.   [2 marks]

_____

_____

_____

Ian and Denise have been referred to a genetic counsellor.

They wish to have their first child soon but are concerned about the risk of having a child with cystic fibrosis, as a relative of Ian's has cystic fibrosis.

Ian and Denise do not suffer from cystic fibrosis.

The genetic counsellor takes DNA samples from Ian and Denise and sends them to a laboratory to determine the probability of them having a child with cystic fibrosis. The results are shown in the DNA fingerprint opposite.

(ii) How does the process of gel electrophoresis produce the DNA fingerprint?   [2 marks]

_____

_____

10

Normal CFTR allele	Mutant CFTR allele	Ian	Denise
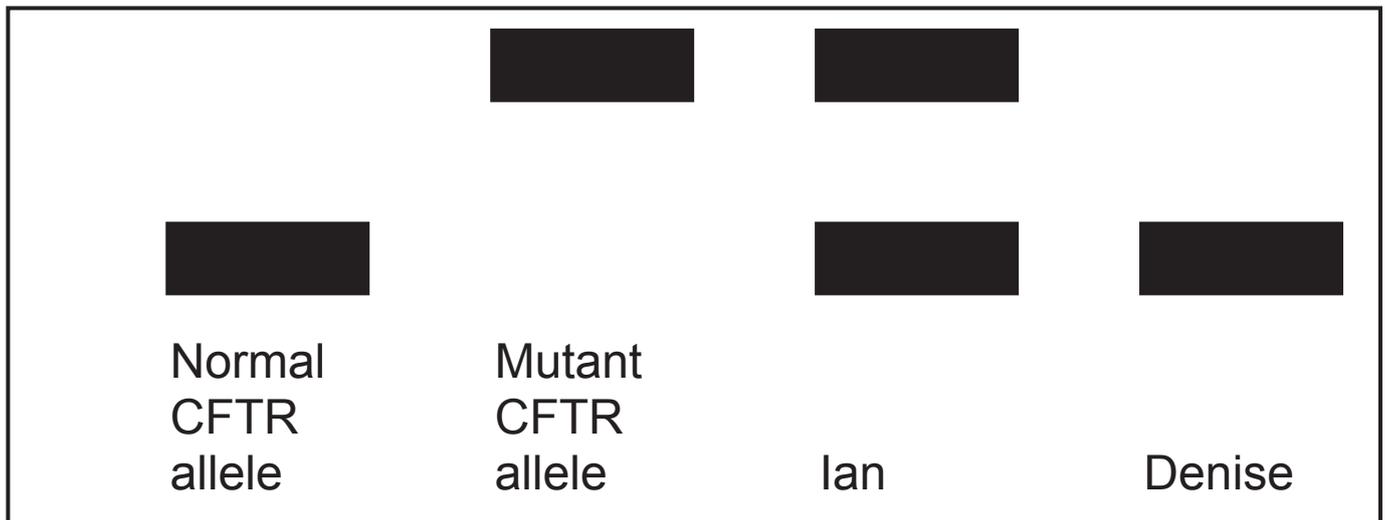
(iii) Using the result of the DNA fingerprint, describe and explain the advice the genetic counsellor would give to Ian and Denise regarding the probability of them having a child with cystic fibrosis. [3 marks]

_____

_____

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_____

(b) The Human Genome Project has identified an increased risk of developing some cancers, such as breast and ovarian cancers, with mutations in certain genes.

Two of these genes with mutations are the **BRCA1** and **BRCA2** genes.

A woman visits a genetic counsellor. Her sister and aunt have been diagnosed with breast cancer.

(i) Suggest the advice a genetic counsellor could give this woman.   [2 marks]
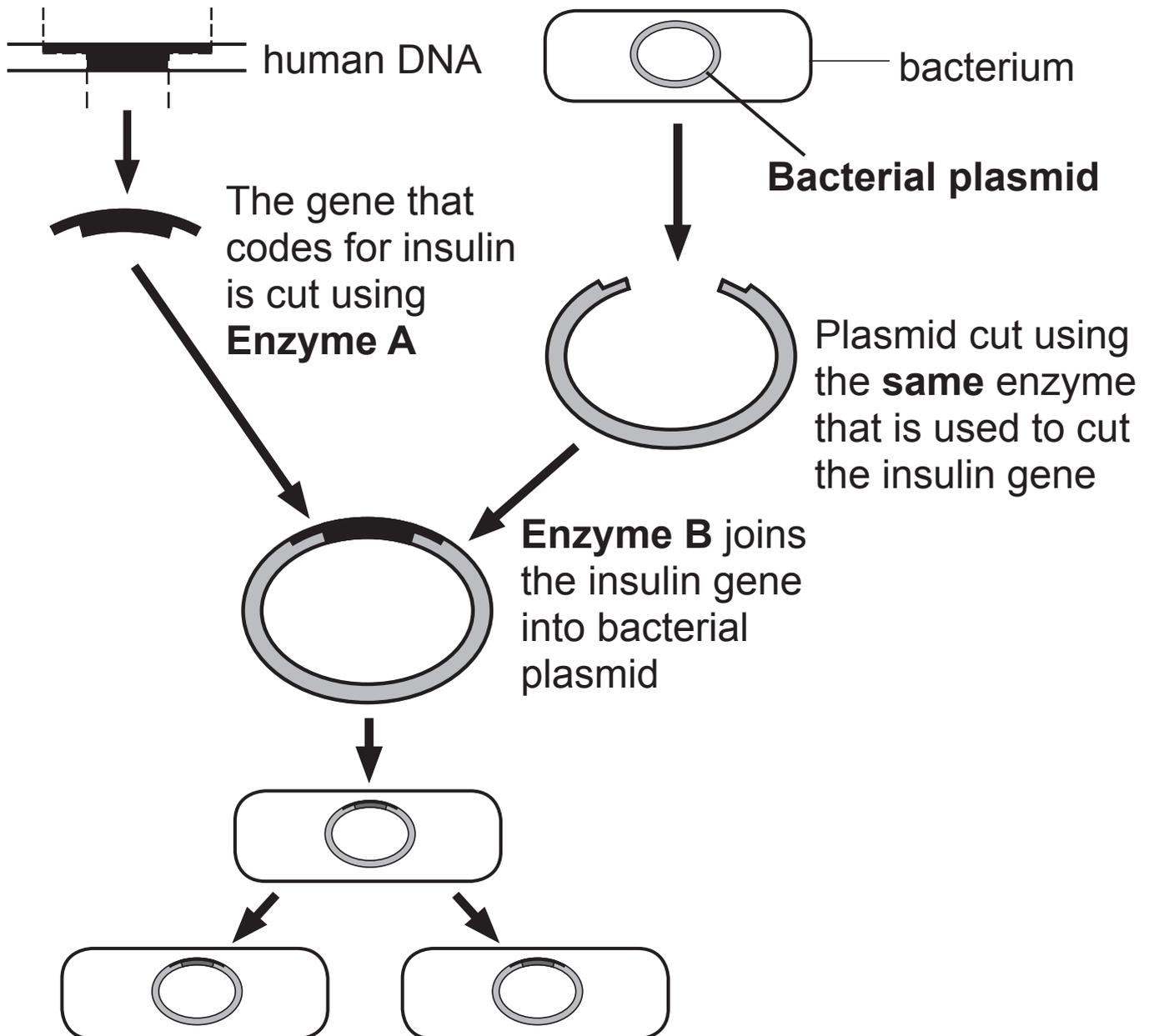
_____

_____

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_____

Another woman was found to have a mutation in the **BRCA1** gene.

(ii) Explain why this result does not mean that this woman will definitely develop breast cancer.
[1 mark]

_____

_____

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(Questions continue overleaf)

[Turn over

5 **(a)** The diagram below shows how insulin can be made using genetically modified bacteria.

human DNA

bacterium

Bacterial plasmid

The gene that codes for insulin is cut using **Enzyme A**

Plasmid cut using the **same** enzyme that is used to cut the insulin gene

Enzyme B joins the insulin gene into bacterial plasmid

(i) Give the names of Enzyme **A** and Enzyme **B**.
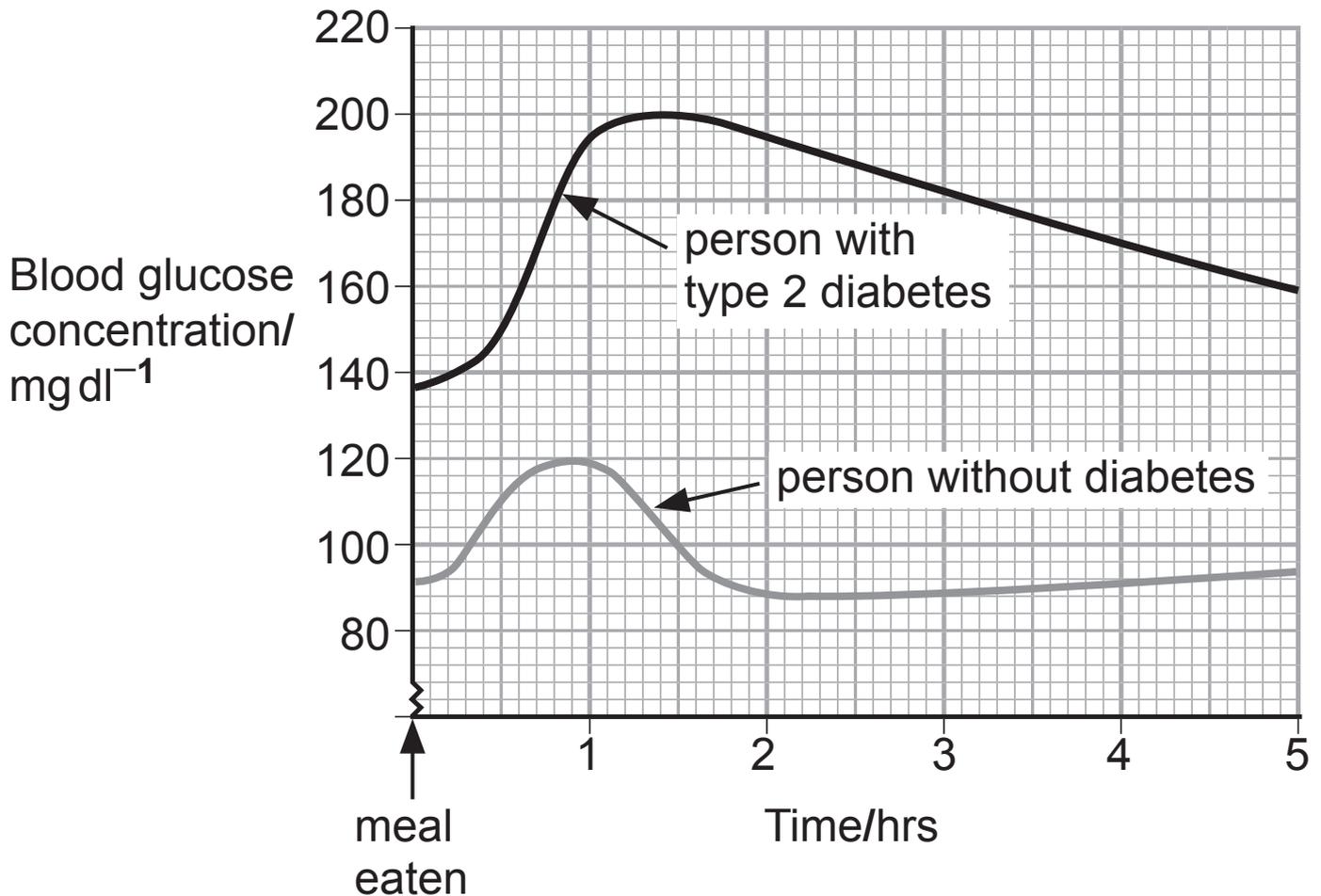[2 marks]

Enzyme **A** _____

Enzyme **B** _____

(ii) The **same** enzyme (Enzyme **A**) is used to cut the insulin gene and the bacterial plasmid. Explain why. [2 marks]

_____

_____

_____

(iii) Name one other medical substance that is produced by genetic engineering.   [1 mark]

_____

15

[Turn over

(b) The graph below shows the blood glucose concentration of a person without diabetes and a person with type 2 diabetes.

Use the graph and your knowledge to: [8 marks]

- describe and explain the changes in blood glucose concentration in a person without diabetes between one and two hours after eating a meal.

- state and describe the difference in blood glucose concentration of a person without diabetes compared to a person with type 2 diabetes over the five hours.

Use data to support your answer.

You will be assessed on the quality of written communication in this question.

11982.02 **MV18**

16

- Describe and explain the changes in blood glucose concentration in a person without diabetes between one and two hours after eating a meal.

_____

_____

_____

_____

_____

_____

_____

_____

- State and describe the difference in blood glucose concentration of a person without diabetes compared to a person with type 2 diabetes over the five hours.

  Use data to support your answer.

_____

_____

_____

_____

_____

_____

_____

6 In squash plants, fruit colour is determined by the interaction of two genes, **W/w** and **G/g**.

The **W** allele suppresses the action of the **G/g** gene so the fruit are white.

When the genotype is **ww**, the **G/g** gene is expressed, the dominant **G** allele determines yellow fruit colour, whilst the recessive **g** allele determines green fruit colour.

(a) (i) Define the term 'gene'.   [2 marks]

_____

_____

(ii) What term is used to describe the interaction between the genes **W/w** and **G/g**?   [1 mark]

_____

(b) Two squash plants of the following genotypes were crossed.

WwGg × **WwGg**

(i) Complete the Punnett square to show the parental gametes and the genotypes of the offspring.
[4 marks]

(ii) Give the numbers of each of the phenotypes produced in this Punnett square.   [3 marks]

White _____

Yellow _____

Green _____

7 Rennet obtained from calves has traditionally been used in cheese-making to coagulate milk. Due to increased demand for this enzyme, several other sources of rennet are being used.

These sources include a rennet enzyme extracted from the fungus **Rhizomucor miehei**.

(a) Scientists carried out an investigation into the effect of temperature and pH on the average milk coagulation time when the rennet of **Rhizomucor miehei** was used.

The results obtained by the scientists are shown in the table below.

	Average milk coagulation time/s	
Temperature/°C	pH 5.0	pH 6.0
30	37	50
35	27	37
40	22	20
45	22	24
50	19	19
55	16	18
60	18	100

(i) Using the information provided, draw a graph showing the effect of temperature and pH on the average milk coagulation time of **Rhizomucor miehei** rennet.   [5 marks]

21

[Turn over
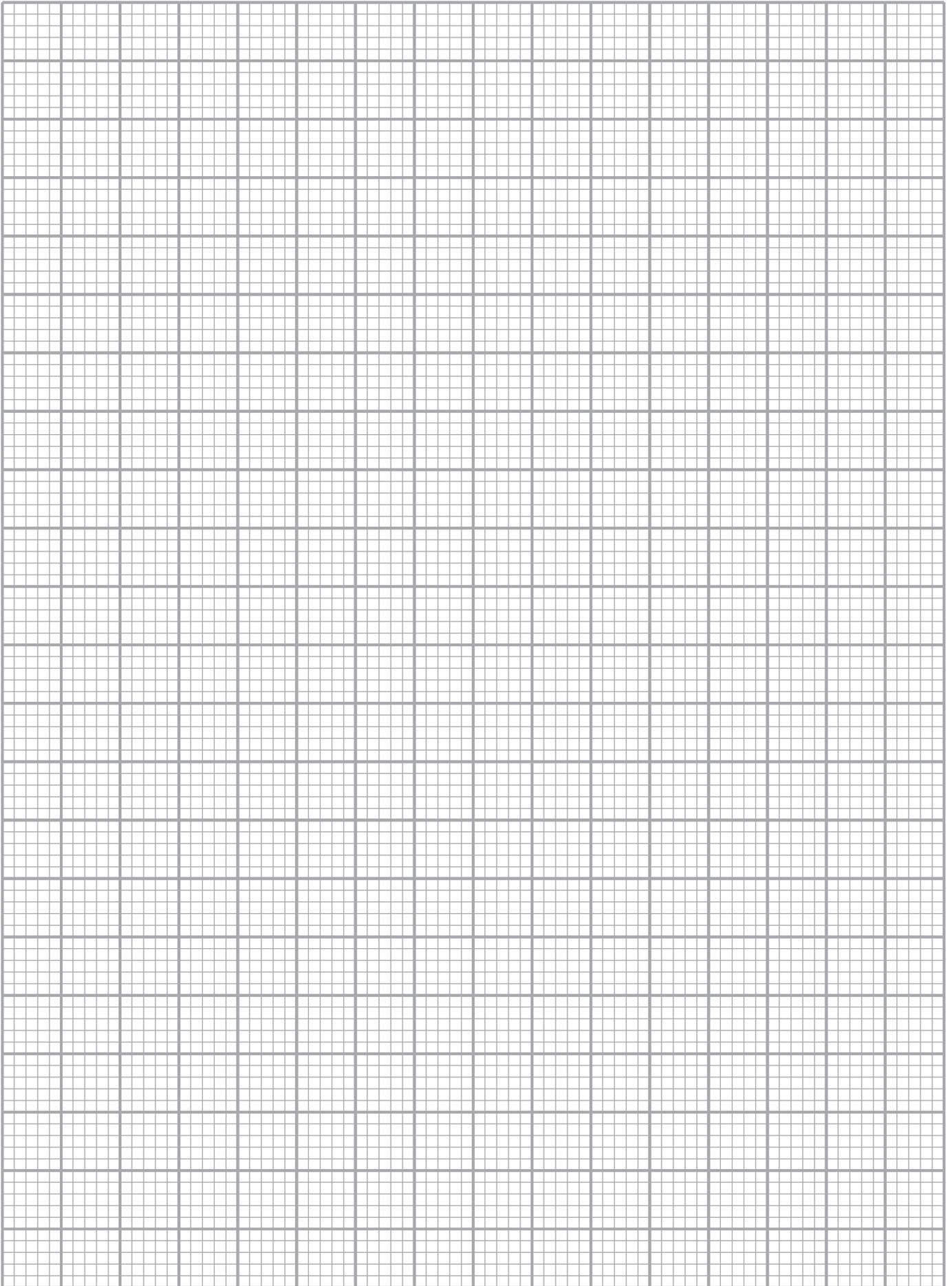
(ii) Determine the **optimum** temperature and pH of **Rhizomucor miehei** rennet in the coagulation of milk.   [2 marks]

Temperature _____ °C

pH _____

(iii) Which temperature shows the greatest difference in milk clotting time between pH 5.0 and 6.0?
[1 mark]

_____ °C

(iv) Suggest **two** variables the scientists controlled to ensure the results were valid.   [2 marks]

1. _____

2. _____

(v) Scientists measured the average milk coagulation time at intervals of 5°C.
How could scientists improve their method to obtain a more exact value for the optimum temperature of milk coagulation?   [2 marks]

_____

_____

_____

Chymosin is an alternative to rennet used in the cheese-making industry to coagulate milk.

(b) Give **two** advantages of using chymosin as an alternative to rennet.   [2 marks]

1. _____

2. _____

8 Gene therapy can be used to treat people with certain genetic conditions.

(a) Explain what is meant by the term 'gene therapy'.
[2 marks]

_____

_____

_____

(b) Give **three** differences between somatic cell therapy and germ line cell therapy.   [3 marks]

1. _____

_____

2. _____

_____

3. _____

_____

(c) Give **two** potential benefits of somatic cell therapy to treat a person suffering from a genetic disease.
[2 marks]

1. _____

_____

2. _____

_____

9 Sickle cell anaemia is a condition caused by a mutation in a gene that codes for haemoglobin.

The gene that controls the production of haemoglobin is located on chromosome 11.

(a) A gene occupies a fixed position on a chromosome.

   (i) Give the term that describes this position.
   [1 mark]

   _____

The gene containing the mutation that causes sickle cell anaemia is 438 bases long.

   (ii) Calculate the number of **amino acids** coded for by this gene.   [2 marks]

   You are advised to show your working.

   _____ amino acids

The alleles of the gene that causes sickle cell anaemia are **co-dominant**.

(iii) Explain what is meant by the term co-dominant. [2 marks]

_____

_____

_____

The normal allele for haemoglobin is Hb^A. The abnormal form that causes sickle cell anaemia is caused by the allele Hb^S.

Individuals who are heterozygous for sickle cell anaemia have the genotype $Hb^A Hb^S$.

These individuals do **not** have sickle cell anaemia but are described as having the sickle cell trait.

(iv) Give the **genotype** of an individual who has sickle cell anaemia. [1 mark]

_____

26

(v) If two people who are heterozygous for sickle cell anaemia ($Hb^A Hb^S$) have a child, determine the probability of them having a child with sickle cell anaemia.   [3 marks]

Show your working in a Punnett square in the space below.

Probability: _____

(b) In a study, the genotype of **500** children living in an African village was recorded:

- 110 are homozygous and do not suffer from sickle cell anaemia

- 279 are heterozygous for sickle cell anaemia

- 111 suffer from sickle cell anaemia

The chi-squared test can be used to check if these results statistically fit an expected ratio of **1:2:1**.

The formula for the chi-squared test is:

$$\chi^2 = \sum \frac{(O - E)^2}{E}$$

where O = observed frequency
E = expected frequency
$\sum$ = sum of.

(i) Complete the table below and then calculate the χ^2 value for these results.   [5 marks]

Category	Observed (O)	Expected (E)	(O – E)	(O – E)^2	$\dfrac{(O - E)^2}{E}$
Homozygous and do not suffer from sickle cell anaemia	110				
Heterozygous for sickle cell anaemia	279				
Suffer from sickle cell anaemia	111				

Calculated χ^2 value _____

The table below gives probability values for use in a χ^2 test with $n-1$ degrees of freedom (d.f.) where n is the number of categories.

χ^2 values

d.f.	probability = 0.900	0.500	0.100	0.050	0.010	0.001
1	0.016	0.455	2.71	3.84	6.63	10.83
2	0.211	1.39	4.61	5.99	9.21	13.82
3	0.584	2.37	6.25	7.81	11.34	16.27
4	1.06	3.36	7.78	9.49	13.28	18.47
5	1.61	4.35	9.24	11.07	15.09	20.52
6	2.20	5.35	10.64	12.59	16.81	22.46
7	2.83	6.35	12.02	14.07	18.48	24.32
8	3.49	7.34	13.36	15.51	20.09	26.13
9	4.17	8.34	14.68	16.92	21.67	27.88

(ii) What is the number of degrees of freedom (d.f.) for this test?   [1 mark]

_____

(iii) Between what **range of probabilities** does your χ^2 value fit?   [1 mark]

Use the table above to assist you.

Between _____ and _____

(iv) Explain fully the outcome of your statistical test.
[2 marks]

_____

_____

_____

_____

_____

(c) Malaria is a disease caused by the parasite **Plasmodium falciparum** which is carried by the female mosquito.

The malaria parasite is less able to complete its life cycle in sickle shaped red blood cells.

The diagram below shows the distribution of **P. falciparum** malaria and sickle cell anaemia in Africa.

P. falciparum malaria   Sickle cell allele

Malaria

<1%

1−5%

5−10%

10−20%
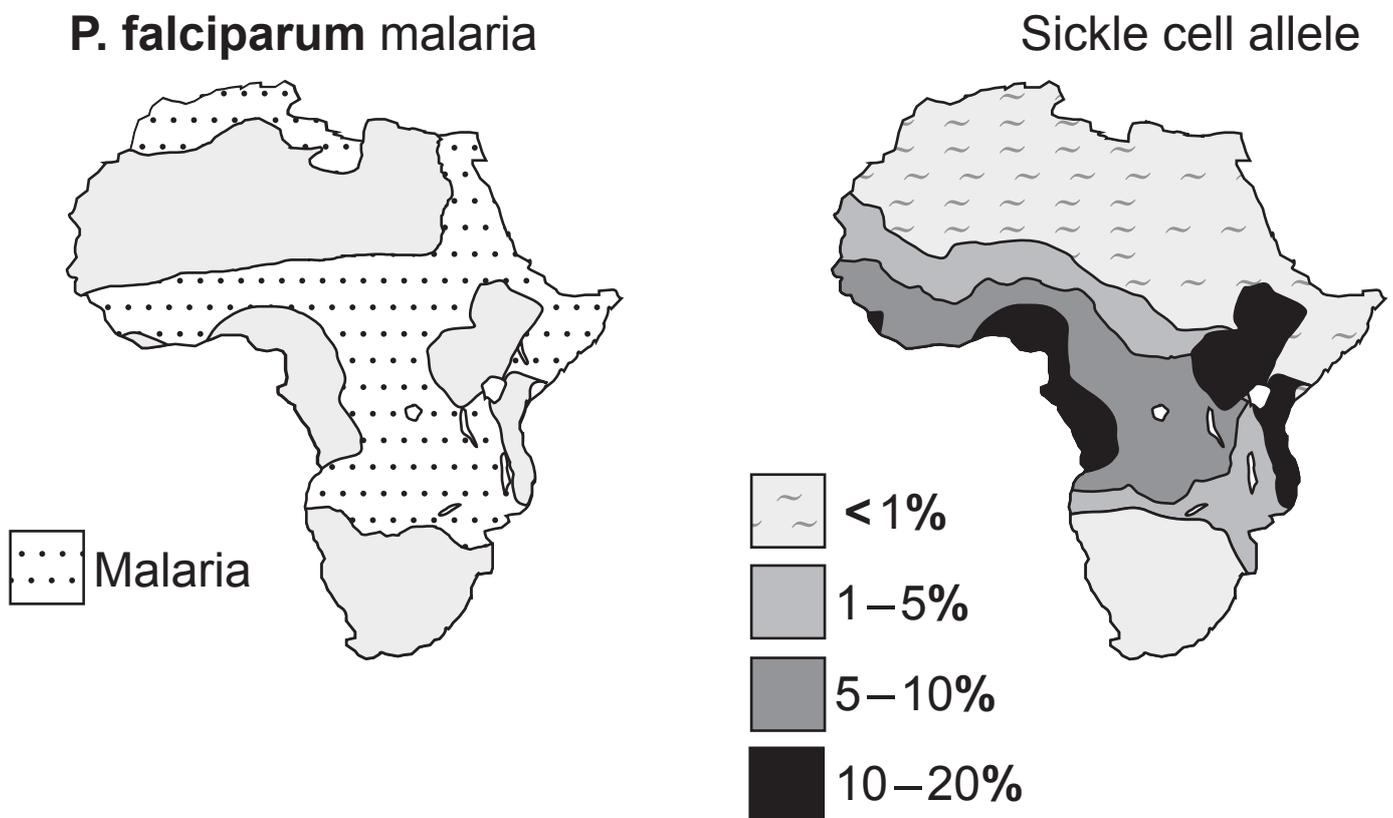
(i) Compare the distribution of malaria to that of sickle cell allele in Africa.   [2 marks]

_____

_____

_____

(ii) Suggest why there is **<1%** incidence of sickle cell anaemia in Southern Africa.   [1 mark]

_____

_____

_____

This is the end of the question paper

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Q9(b)(i) . . Source: *CCEA*

Q9(c) . . . Source: *Principal Examiner*

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Question Number	**Marks**
1	
2	
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Total Marks	

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