Rewarding Learning

ADVANCED
General Certificate of Education
2024

Centre Number

Candidate Number

Life and Health Sciences

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

AZ051

[AZ051]
TUESDAY 18 JUNE, MORNING

TIME

1 hour 45 minutes.

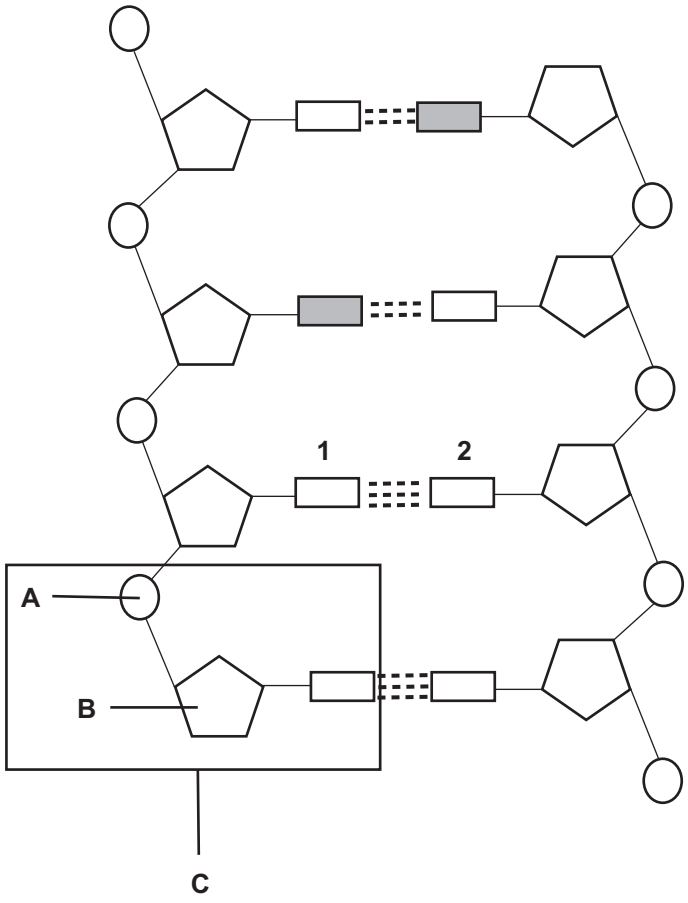
INSTRUCTIONS TO CANDIDATES

Write your Centre Number and Candidate Number in the spaces provided at the top of this page.
You must answer the questions in the spaces provided.
Do not write outside the boxed area on each page or on blank pages.
Complete in black ink only. **Do not write with a gel pen.**
Answer **all nine** questions.

INFORMATION FOR CANDIDATES

The total mark for this paper is **100**.
Figures in brackets printed down the right-hand side of pages indicate the marks awarded to each question or part question.
You may use an electronic calculator.
Quality of written communication will be assessed in question **6(a)**.

1 The diagram below shows a short section of a nucleic acid.



(a) State what the dotted lines shown in the above diagram represent.

_____ [1]

(b) Name the components labelled **A**, **B** and **C**.

A _____

B _____

C _____ [3]

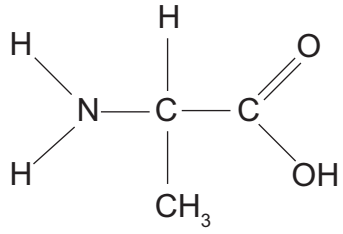
(c) The shaded bases in the diagram represent adenine (A).

Suggest the names of the bases labelled **1** and **2**.

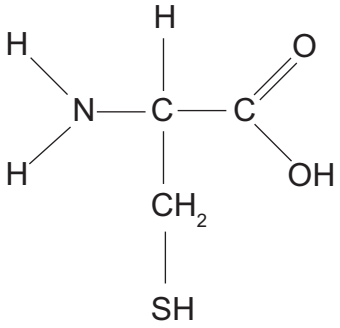
_____ and _____ [1]

[Turn over

2 Two different amino acids are shown in the diagram below.



Alanine



Cysteine

(a) Give **one** similarity and **one** difference in the structures shown above.

Similarity _____

_____

Difference _____

_____ [2]

(b) Mutations are changes that can occur in genes.

There are several types of mutations.

Name **three** types of gene mutations.

1. _____

2. _____

3. _____ [3]

(c) Genes are sections of DNA coding for proteins.

(i) Define the term **allele**.

_____

_____ [1]

There are some traits (characteristics) that are coded for by multiple alleles.

There are four blood groups, A, B, AB and O.

The genotype for blood group O is I^o I^o, the alleles are O and O.

The alleles A and B are codominant.

The alleles A and B are dominant to the allele O.

(ii) State the possible alleles that may be present in an individual with blood group A.

_____

_____ [2]

(iii) An individual has blood group AB.

Give the genotype of this individual.

_____ [1]

[Turn over

3 DNA probes can be used to diagnose medical conditions such as some types of breast cancers.

(a) (i) Define the term **DNA probe**.

_____

_____

_____

_____ [2]

(ii) Explain why the sequence of the human genome needed to be known before probes could be used to locate disease-causing genes.

_____

_____

_____

_____

_____ [2]

(iii) DNA probes can be labelled in different ways.

Originally, they were radioactively labelled, they are now more commonly labelled with chemicals which are fluorescent.

Give **one** advantage of using fluorescent chemicals to label probes.

_____

_____

_____ [1]

(iv) Suggest why some genetic diseases cannot be tested for using a DNA probe to a sequence on **only one** gene.

_____

_____

_____

_____ [1]

(b) Individuals who have been diagnosed with a genetic disease will often be referred to a genetic counsellor.

(i) Explain the role of the genetic counsellor in this process.

_____

_____

_____

_____

_____

_____ [2]

(ii) Suggest **three** possible considerations the individual may have during and after the counselling process.

1. _____

_____

2. _____

_____

3. _____

_____ [3]

[Turn over

(iii) Suggest a reason why not all individuals with a disease-causing gene go on to develop the disease.

_____

_____ [1]

4 The polymerase chain reaction (PCR) is used to make many copies of (amplify) small samples of DNA in a short period of time.

The amount of DNA doubles with every cycle.

(a) (i) Suggest **one** example when it is useful to produce many copies of a DNA sample.

_____

_____ [1]

(ii) A PCR machine completes 15 cycles (doubling the number of DNA molecules in each cycle).

If the original sample contained 1 molecule of DNA this would produce 32 768 (3.3×10^4) molecules of DNA.

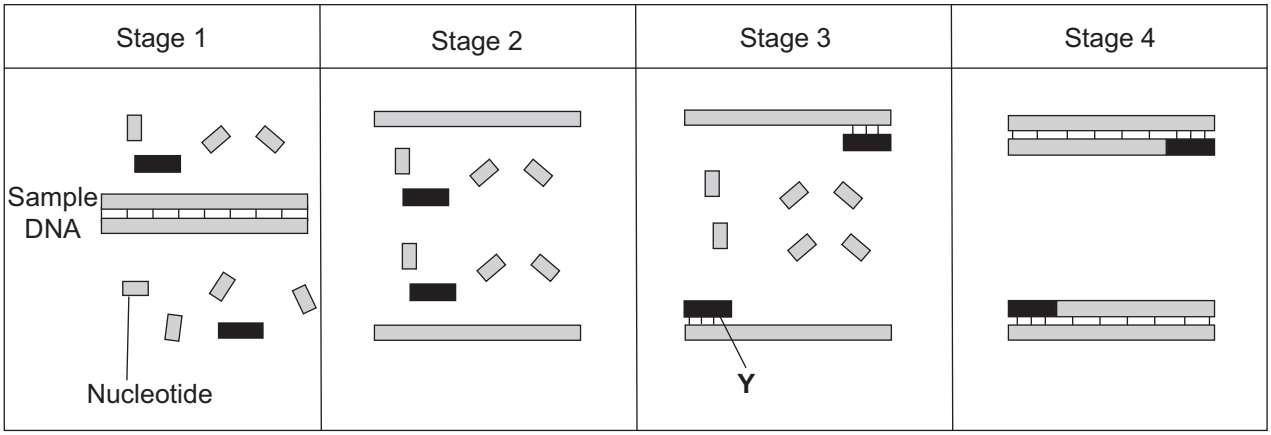
How many molecules would be produced in total if the machine completed **one more** cycle?

Show your working out.

_____ [2]

[Turn over

The diagram below represents some stages of the PCR cycle.



(b) (i) State the temperature required for stage 2.

_____ °C [1]

(ii) State what has happened to the DNA sample by stage 2.

_____

_____

_____ [2]

(iii) Identify and explain **two** of the roles played by the molecule labelled **Y** in stages 3 and 4.

Y: _____

1. _____

_____

2. _____

_____ [3]

(iv) In 1987 a new enzyme needed for stage 4 was isolated from heat-loving bacteria (>95°C).

This is now the standard enzyme used for PCR.

Suggest **one** benefit of using this enzyme in the process of PCR.

_____

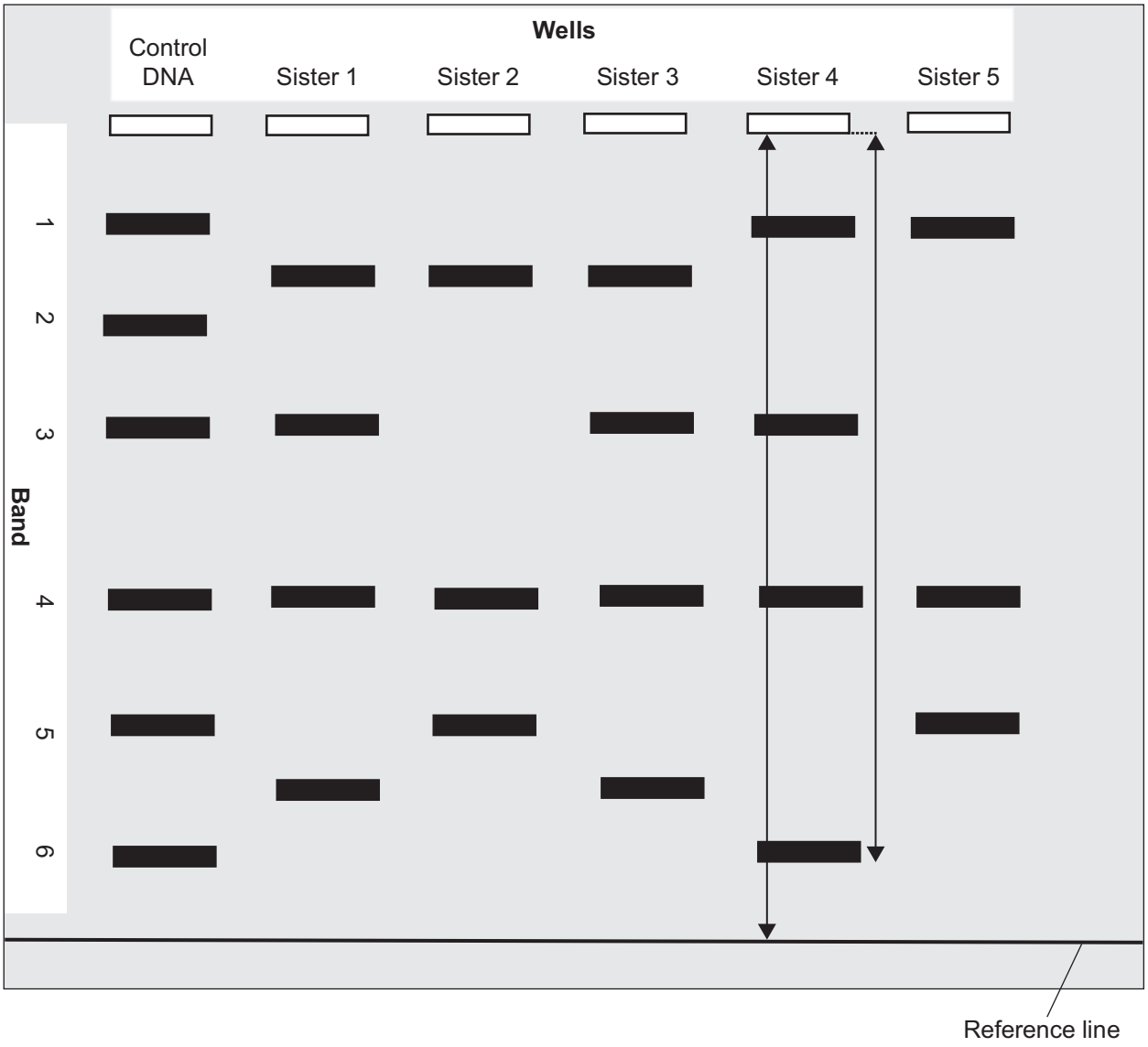
_____

_____

_____ [1]

[Turn over

5 The diagram below represents a gel electrophoresis used to determine the risk of developing breast cancer due to a mutation of the *BRCA2* gene amongst a family of sisters (1–5).



(a) Describe how the process of gel electrophoresis was carried out to produce the results shown in the diagram.

[5]

[Turn over

(b) (i) State how many restriction sites are present in the DNA of **sister 2**.

_____ [1]

(ii) Using the formula for retention factor (R_f):

$$R_f = \frac{\textit{Distance migrated from well by band}}{\textit{Distance from well to reference line}}$$

Calculate the R_f value for band 6 present in **sister 4** (using the arrowed lines shown on the diagram).

Show your working out.

_____ [2]

(iii) Sisters **1** and **4** have already developed breast cancer due to the presence of the *BRCA2* gene.

The others have not shown any symptoms of the disease.

Identify one other sister who may be at risk of developing the disease, giving reasons for your choice.

Sister _____

Reasons _____

_____

_____

_____ [3]

6 DNA replication is a critical part of the process of cell division.

(a) Describe in detail the process of semi-conservative DNA replication.

Quality of written communication will be assessed in this question.

[6]

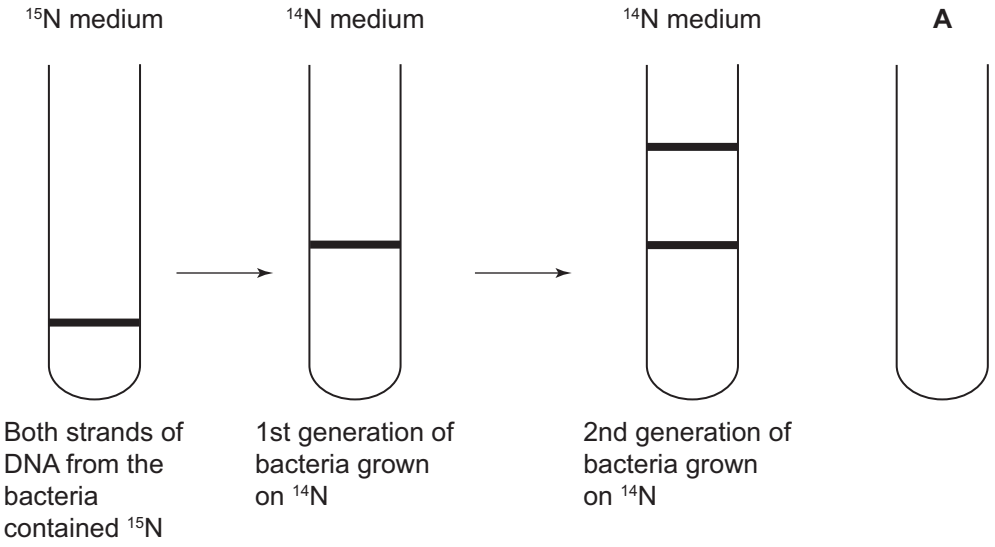
[Turn over

(b) Originally three different theories were suggested to explain DNA replication.

Meselson and Stahl developed an experiment to test which of these theories was correct using two different isotopes of nitrogen (^{14}N and ^{15}N).

^{15}N is a heavier isotope than ^{14}N.

The diagram below shows some of the experimental results they obtained.



(i) In the tube labelled **A**, complete the diagram for the 2nd generation of the **fragmentation theory** (dispersive theory) of DNA replication. [2]

(ii) Briefly outline the main difference between the semi-conservative and the conservative theories of DNA replication.

_____

_____

_____

_____

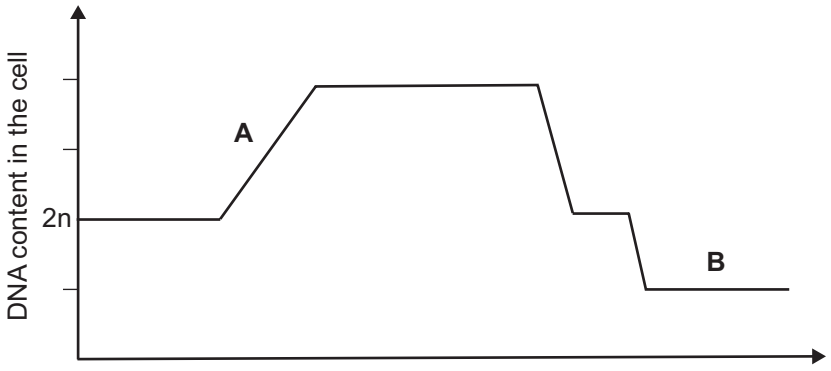
_____

_____ [2]

(c) Although the process of DNA replication produces identical, new double stranded DNA molecules, the cells which finally become gametes have genetic differences.

These genetic differences are produced during the process of meiosis.

The graph below outlines how the DNA content in the cell changes during DNA replication and the stages of meiosis.



(i) State what is happening during the part of the graph labelled **A**.

_____

_____ [1]

(ii) State the number of chromosomes present in a human cell at the region labelled **B** on the graph.

_____ [1]

(iii) Explain how recombination (cross-over) can result in variation within gametes.

_____

_____

_____

_____

_____ [2]

[Turn over

7 **Antitrypsin enzyme** that controls the activity of elastase in the lungs, is normally produced in the liver.

Gene technology involving several techniques can be used to obtain antitrypsin enzyme.

The technology involves transferring the gene for antitrypsin enzyme (target gene) into a fertilised sheep's egg (host cell).

The fertilised egg can then be carried by a surrogate female sheep.

The lamb produced is considered transgenic.

(a) (i) Define the term **transgenic**.

_____

_____

_____ [2]

(ii) Suggest which cells would be a good source of the antitrypsin gene.

_____ [1]

Plasmids are small circular DNA molecules that may be used to transfer the target gene to the host cell, using a microinjector.

(b) (i) State the term used to describe a plasmid acting in this role.

_____ [1]

(ii) Why is it useful to use the same restriction enzyme to obtain the gene **and** cut the plasmid open before inserting the gene?

_____

_____

_____ [1]

(iii) Where in the fertilised egg cell is the modified plasmid inserted using a microinjector?

_____ [1]

(iv) State **two** advantages of producing the antitrypsin enzyme by genetic engineering.

1. _____

_____

_____

2. _____

_____

_____ [2]

(v) In these transgenic sheep, the antitrypsin is produced in their milk.

Suggest a benefit of this method of production.

_____

_____ [1]

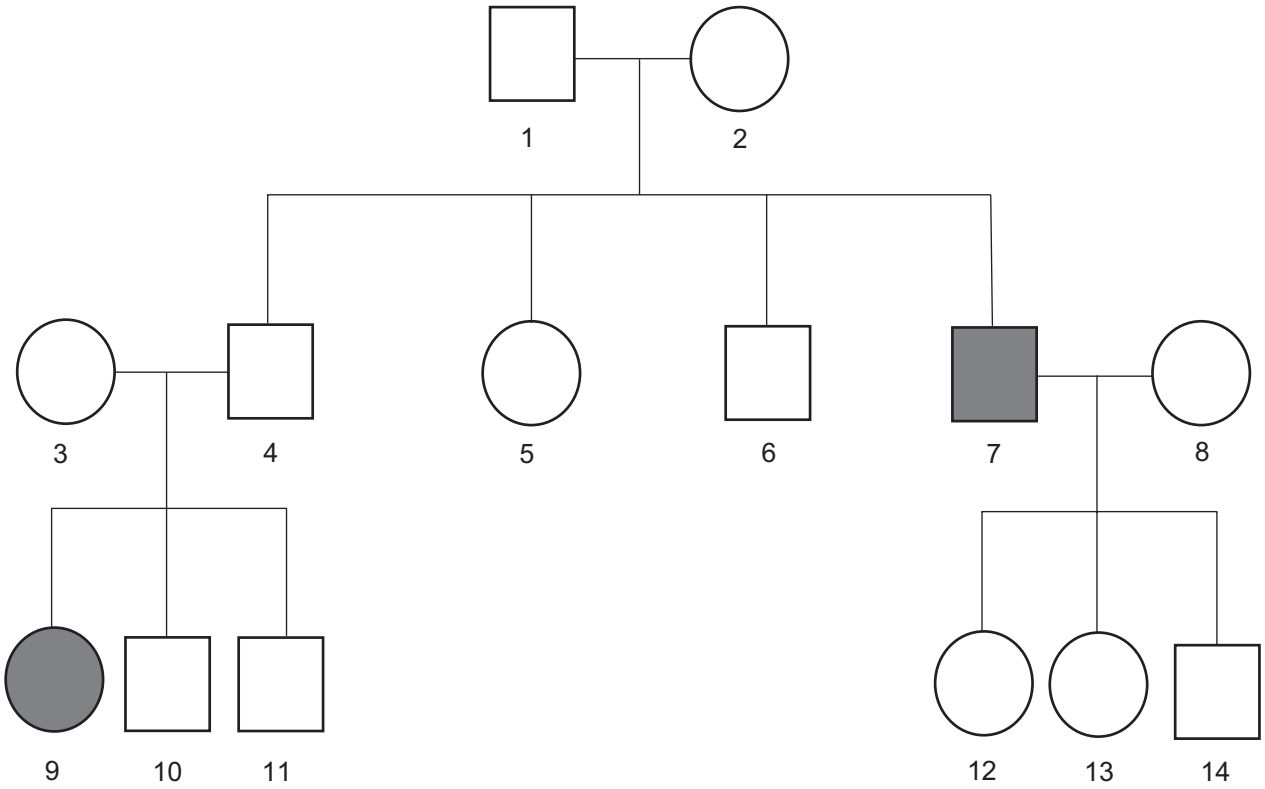
[Turn over

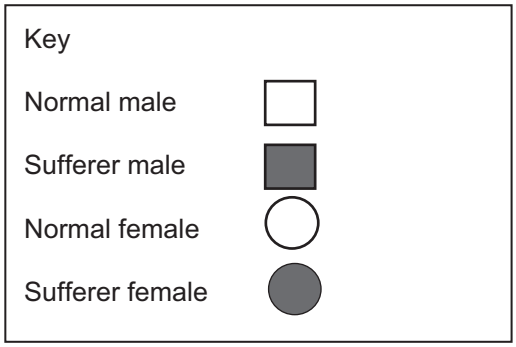
8 There are several inherited genetic disorders that can cause blood clotting problems.

Haemophilia is one, Factor XIII deficiency is another.

Unlike haemophilia, Factor XIII deficiency is **not sex-linked**.

The pedigree diagram below shows the inheritance of Factor XIII deficiency.





(a) (i) Using the information in the pedigree diagram and your knowledge, explain why the relationship between individuals suggests the inheritance of Factor XIII deficiency is:

- **not** sex-linked.

- **not** dominant.

[4]

(ii) Explain what is meant by the term **recessive**.

[1]

[Turn over

(iii) Using the letter **A** (normal allele) and **a** (recessive allele), state the possible genotypes of the following individuals:

Individual 3 _____

Individual 8 _____ [2]

(b) (i) Using the symbols **A** for normal blood and **a** for Factor XIII deficiency, complete the Punnett square below.

Parental phenotypes: normal male × normal female

Parental genotypes: **Aa** **Aa**

[3]

(ii) Circle any individual who may suffer from Factor XIII deficiency. [1]

(iii) Give the probability of an individual having Factor XIII deficiency.

_____ [1]

(c) Stem cells have been recognised as an extremely important way to treat many medical conditions.

(i) Complete the sentence about stem cells.

A stem cell is an _____ cell that may give rise to several different types of cell. [1]

(ii) Explain how a stem cell may give rise to several different types of cell.

_____

_____

_____

_____ [2]

(iii) Suggest why stem cell transplants from healthy donors may be useful in treating a disease.

_____

_____

_____

_____ [2]

[Turn over

9 In tomato plants, two genes control the production of purple-coloured stems and purple-coloured leaves.

For the tomato plant to be coloured purple, at least one dominant allele must be present **at each locus**.

In the heterozygous form, the genotype and phenotype are as follows:

- HhBb purple-coloured stem and purple-coloured leaves.

In the homozygous form, the genotypes and phenotypes are as follows:

- hhBB green-coloured stem and green-coloured leaves.
- HHbb green-coloured stem and green-coloured leaves.

(a) Two heterozygous purple plants were crossed.

The following results were observed.

Parental phenotype: purple-coloured stem and purple-coloured leaves × purple-coloured stem and purple-coloured leaves

Parental genotype: HhBb × HhBb

	HB	Hb	hB	hb
HB	HHBB	HHBb	HhBB	HhBb
Hb	HHBb	HHbb	HhBb	Hhbb
hB	HhBB	HhBb	hhBB	hhBb
hb	HhBb	Hhbb	hhBb	hhbb

State the phenotypes of the offspring and the number of each.

Phenotype	Number

[3]

[Turn over

(b) A different experiment was carried out with two pure breeding tomato plants.

One had purple flowers and long pollen grains.
The other had red flowers and round pollen grains.

The offspring (1st generation, F1) plants were all heterozygous and had purple flowers and long pollen grains.

The F1 plants were crossed to produce the F2 generation.

Normally such a cross would produce offspring in the ratio 9:3:3:1. However, the results in the table below were obtained, showing an approximate 3:1 ratio.

A χ^2 test was carried out to indicate whether the alleles for flower colour and pollen grain shape were linked on the same chromosome.

When the crosses were carried out, the flower colour and pollen grain shapes from 1600 plants were recorded.

The table shows the results of these crosses.
It also shows a partial calculation of the χ^2 value.

(i) Complete the table and calculate the χ^2 value.

Phenotype	**Observed (O)**	**Expected (E)**	**O–E**	**$(O–E)^2$**	**$\frac{(O–E)^2}{E}$**
Purple flowers / long pollen grains	1225	1200	25	625	0.52
Red flowers / round pollen grains	375	400			

Calculated χ^2 value _____ [4]

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

_____ [1]

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

Source: CCEA

(iii) Between what **range of probabilities** does your χ^2 value fit? Use the table **above** to assist you.

between _____ and _____ .

Do the results statistically fit the expected ratio of 3:1? _____

Explain your answer.

_____

_____

_____ [3]

(iv) Explain what the results mean about the relationship between the locations of the two genes.

_____

_____

_____

_____

_____ [2]

THIS IS THE END OF THE QUESTION PAPER

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For Examiner's use only	
Question Number	**Marks**
1	
2	
3	
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Total Marks	

Examiner Number