Rewarding Learning

ADVANCED
General Certificate of Education
2023

Centre Number

Candidate Number

Life and Health Sciences

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

AZ051

[AZ051]
MONDAY 26 JUNE, AFTERNOON

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.
Write your answers in the spaces provided in this question paper.

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)**.

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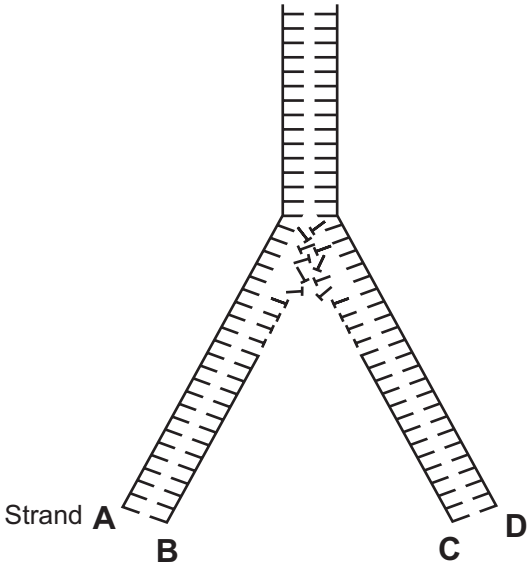
1 The diagram below shows the process of DNA replication in cells.



Complete the sentences below about DNA replication by writing the most appropriate word(s) or letters in the blank spaces.

The diagram shows the parental DNA being separated (unwound) as a

result of the enzyme _____ breaking the

_____ bonds which link complementary bases together.

Two strands then act as templates to form new DNA.

The template strands in the diagram are labelled with the letters

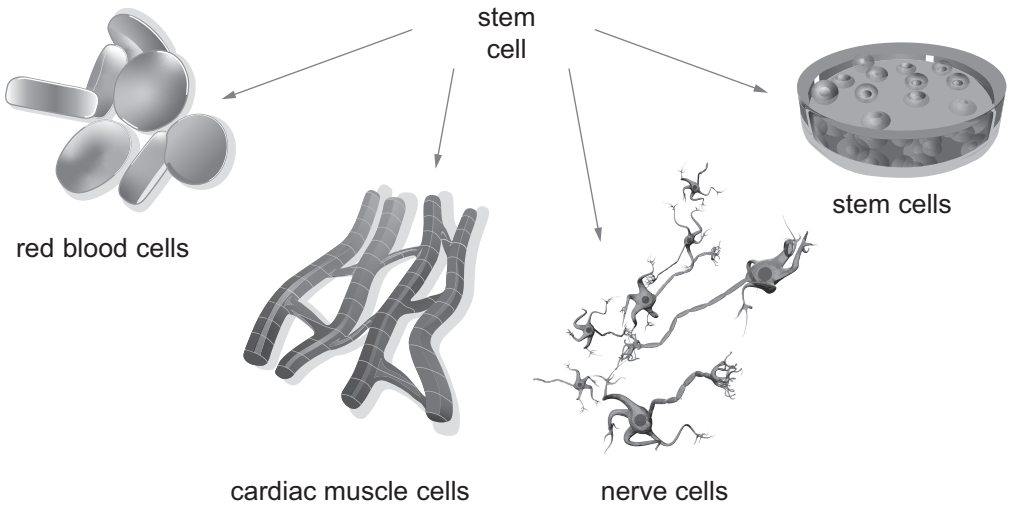
_____ and _____ .

The diagram shows the _____ theory of DNA replication.

[4]

[Turn over

2 The diagram below summarises the properties of stem cells.



Source: © CCEA

(a) Embryonic and adult stem cells are the two main types of stem cell.

(i) Give **one** similarity between embryonic and adult stem cells.

_____

_____ [1]

(ii) Give **one** difference between embryonic and adult stem cells.

_____

_____ [1]

(iii) Name **one** place in the body in which adult stem cells are found.

_____ [1]

(b) Stem cells are being increasingly used in medicine.
Explain why there are few ethical issues about the use of adult stem cells in this role.

_____

_____ [1]

[Turn over

3 Making cheeses such as Cheddar traditionally involves adding protease enzymes to milk.

One of these enzymes is rennet.

It is produced by the stomach lining of young calves.

(a) Give the function of rennet in making cheese.

_____

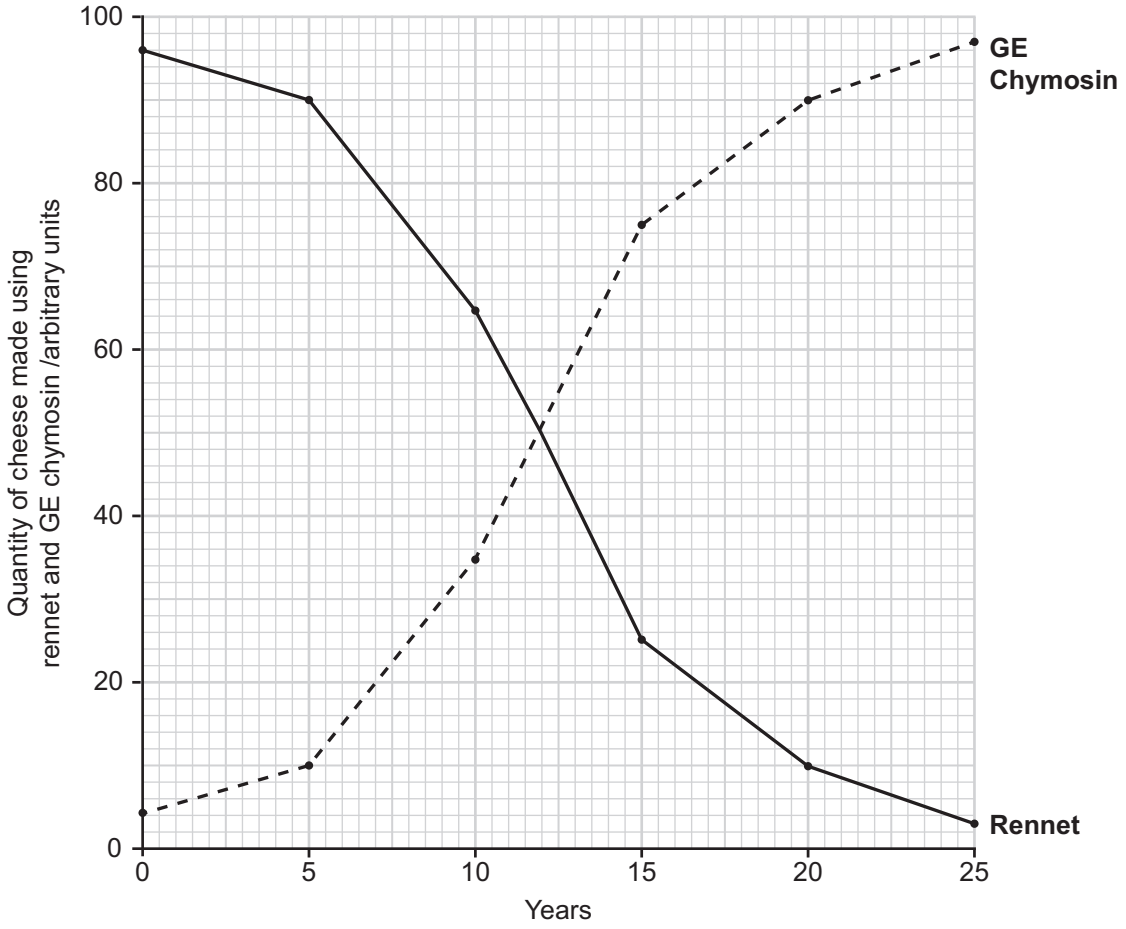
_____ [1]

(b) One protease enzyme, called chymosin, can be used instead of rennet in cheese making.

This enzyme can be produced by genetic engineering (GE) where the gene for chymosin is inserted into bacteria such as *E. coli.*

These modified bacteria then produce large quantities of GE chymosin at low cost.

The graph opposite shows the quantities of cheese made using rennet and GE chymosin over a 25-year period in a large cheese-making company.



(i) After how many years was the quantity of cheese made using rennet and GE chymosin the same?

_____ years [1]

(ii) Calculate the percentage decrease in quantity of rennet used over the 25 years.
You are advised to show your working.

_____ % [3]

[Turn over

(iii) Using the information provided and your knowledge, give three reasons for this decrease.

1. _____

_____

2. _____

_____

3. _____

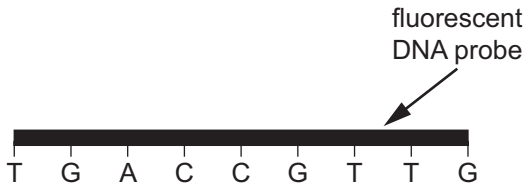
_____ [3]

(iv) Suggest why some manufacturers still use rennet to produce their cheese.

_____ [1]

(c) During genetic engineering, DNA probes are often used to identify the position of a gene.

The diagram below shows part of a DNA probe which is labelled with a fluorescent substance.



(i) Using the diagram and your knowledge, explain how a DNA probe can be used to identify the location of a gene of interest.

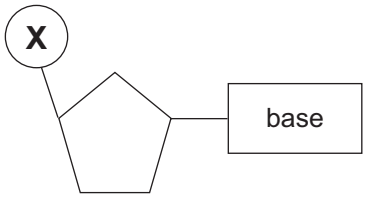
[3]

(ii) DNA probes can also be used to screen medical patients.
Give **one** example of DNA probes in this context.

[1]

[Turn over

4 The diagram below shows the structure of a DNA nucleotide.



(a) Name the component labelled **X**.

_____ [1]

(b) By knowing the percentage of any one of the four bases in a section of DNA, it is possible to work out the percentages of the other bases present.

(i) Complete the table below by calculating the percentage of the remaining bases.

Name of base	Percentage of base in a section of DNA
Adenine	28
Guanine	
Cytosine	
Thymine	

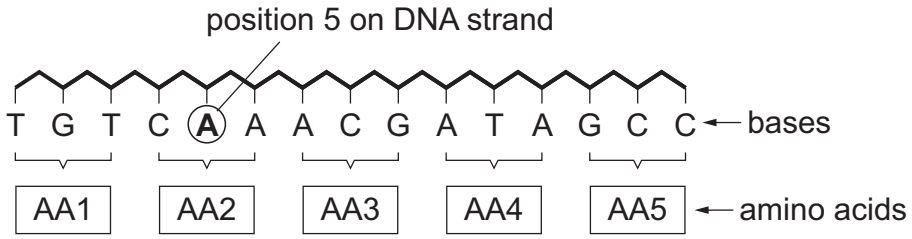
[3]

The relationship between the relative proportions of the different bases is referred to as base equivalence.

(ii) Name the scientist who is credited with working out the principle of base equivalence.

_____ [1]

(c) The diagram below shows a sequence of bases along the coding strand of DNA. This determines the order in which amino acids are built up in a polypeptide.



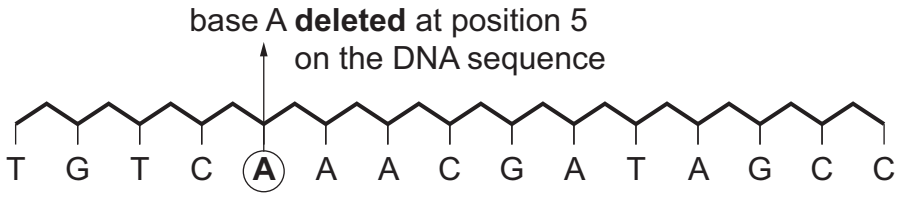
Occasionally, DNA can become damaged by one or more nucleotides being added or deleted from a section of DNA.

(i) State the name given to a change in a DNA sequence.

_____ [1]

[Turn over

The diagram below shows the same DNA sequence. The base at position 5 becomes deleted.



(ii) Describe the effect deleting base A at position 5 will have on the polypeptide produced.

[4]

The original section of DNA could have changed in another way.

The **A** at position 5 on the DNA sequence could have been replaced with a different base rather than being deleted.

(iii) Describe how this would affect the polypeptide produced.

_____

_____

_____

_____

_____ [2]

[Turn over

5 Insulin is a hormone which lowers blood glucose levels.

(a) State **one** way in which insulin lowers blood glucose levels.

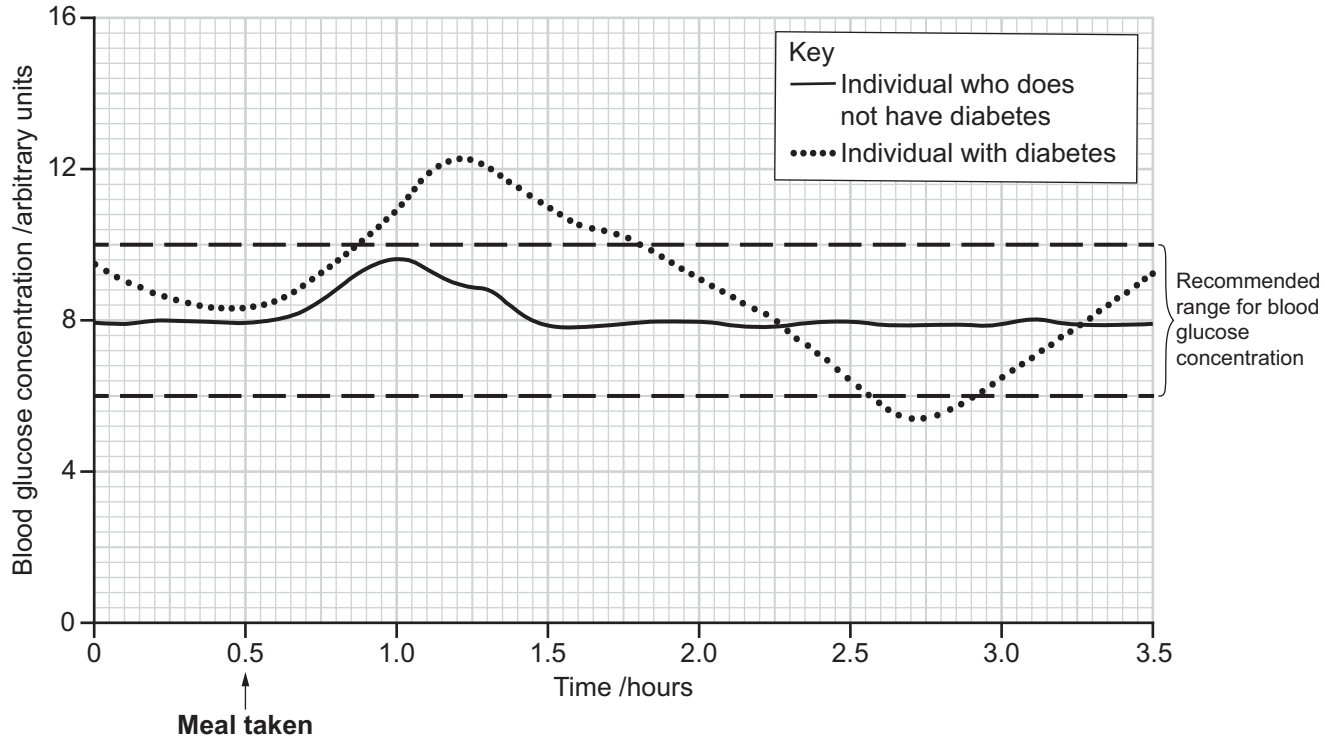
_____

_____ [1]

Diabetes mellitus is a condition in which affected individuals do not produce enough (or any) insulin, or the insulin produced does not work effectively.

The graph below shows the blood glucose levels of two individuals, one with diabetes and one who does not have diabetes, over a period of several hours.

Both had a similar meal at time 0.5 hours.



(b) Compare and contrast the blood glucose concentrations for the two individuals over the period shown.

[6]

[Turn over

(c) As part of the process of producing genetically engineered insulin, it was necessary to isolate the human insulin gene.

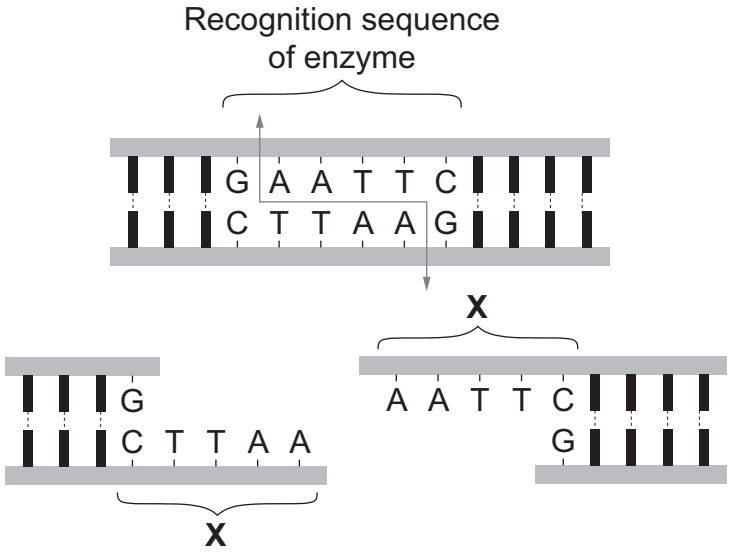
(i) State the name given to the position of a gene on a section of DNA.

_____ [1]

Once the location of the gene was identified, special enzymes were used to cut the DNA on either side of the gene.

These enzymes recognise a series of bases in DNA (a recognition sequence).

In the example shown below, the enzyme recognises the sequence GAATTC and then cuts the DNA between adjacent A and G bases as shown.



Source: Adapted from page 193, © Biology for CCEA A2 Level, 2nd Edition (2017), Dr James Napier, publisher Colourpoint Educational, ISBN: 978-1780-731001

(ii) Name the type of enzymes involved in cutting the DNA.

_____ [1]

(iii) Name the sections of DNA labelled **X**.

_____ [1]

Once the insulin gene has been isolated, it is important that it is then inserted within the bacterial DNA in a certain place so that it will lead to the bacteria producing the insulin as required.

(iv) Explain how the sections of DNA labelled **X** can help with this process.

_____

_____

_____

_____ [2]

[Turn over

(d) People with haemophilia are unable to produce all the chemicals required to clot their blood.

They are unable to produce factor VIII.

(i) State why individuals with haemophilia are unable to produce factor VIII.

_____ [1]

Factor VIII can now be produced by genetic engineering and used to treat people with haemophilia.

(ii) State how people with haemophilia were treated before genetically engineered factor VIII became available.

_____

_____ [1]

(iii) Give **one** health advantage of using genetically engineered factor VIII rather than the earlier treatment method.

_____

_____ [1]

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(Questions continue overleaf)

[Turn over

6 Genetic fingerprinting is a technique increasingly used in forensic science, plant and animal breeding and medical diagnosis.

(a) Describe the process of genetic fingerprinting.

Your answer should include:

- the role of the polymerase chain reaction (PCR) and why it may be necessary;
- the parts of the genome involved and their significance; and
- the laboratory techniques involved in producing the fingerprint.

Quality of written communication will be assessed in this question.

[8]

(b) Give **one** reason for the use of genetic fingerprinting in medical diagnosis and in animal breeding.

Medical diagnosis

Animal breeding

[2]

[Turn over

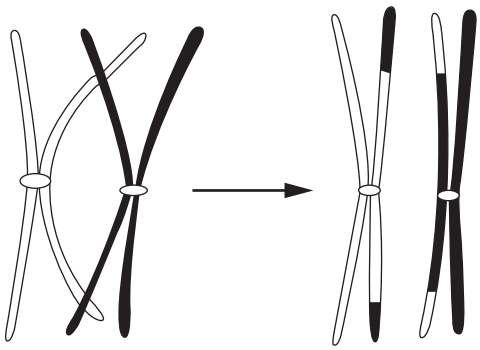
7 Meiosis is a type of cell division often described as 'reduction' division.

(a) Using your knowledge of meiosis, suggest why the term 'reduction' is used.

_____

_____ [1]

(b) The diagram below shows a pair of homologous chromosomes during an early stage (prophase 1) of meiosis.



Source: Principal Examiner

(i) Name the process represented by the diagram.

_____ [1]

(ii) How many chiasmata are shown?

_____ [1]

(iii) Using the diagram and your knowledge, explain the significance of this process.

_____

_____

_____

_____

_____

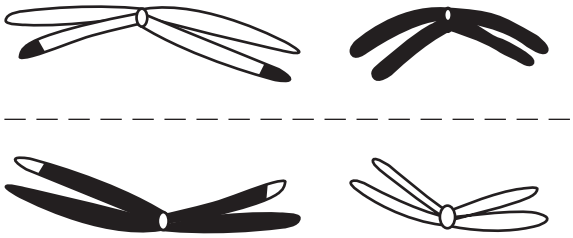
_____ [3]

(c) Read the passage below and answer the questions which follow.

Following the stage of meiosis in **(b)**, each pair of homologous chromosomes line up at the equator in a random arrangement.

This diagram shows two pairs of homologous chromosomes.

When they have lined up at the equator, each pair of homologous chromosomes gets pulled apart to the two different poles of the cell.



Source: Principal Examiner

(i) Name the process by which chromosomes line up at the equator.

_____ [1]

(ii) Explain how this process contributes to variation.

_____

_____

_____

_____

_____ [2]

[Turn over

(d) Down's Syndrome is a condition where an extra chromosome is present in the body cells of the individual.

(i) Give the number of chromosomes in a human **gamete** that has an extra chromosome.

_____ [1]

(ii) Using your knowledge of meiosis, suggest how a gamete could be produced which has an extra chromosome.

_____

_____

_____ [1]

(e) The relationship between the number of babies born with Down's Syndrome in Northern Ireland and the maternal age when the baby is born is shown in the table below.

Maternal age	Number of children born with Down's Syndrome (rate per 1000 total births)
19 and under	0.67
20 – 24	0.85
25 – 29	0.76
30 – 34	1.28
35 – 39	4.50
40 – 44	15.33
45 and over	21.93

Adapted from Table 14b Rate Down's Syndrome births, maternal age, Northern Ireland 2012–2016. NISRA

(i) Summarise the data shown in the table.

[3]

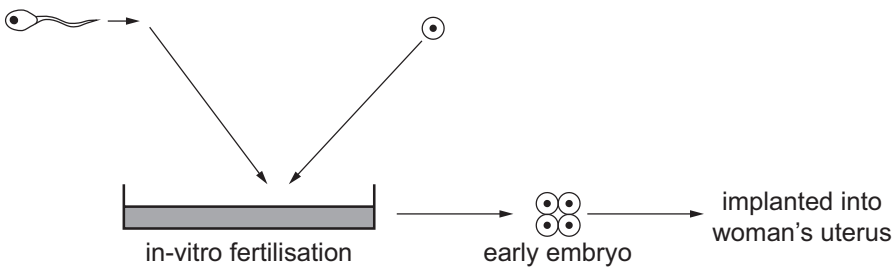
(ii) Suggest a possible explanation for the results shown.

[1]

[Turn over

8 **(a)** The diagram below shows the process of 'in-vitro' fertilisation that can be used during fertility treatment.

If fertilisation takes place, the resulting embryo is implanted into the woman's uterus.



Gene therapy is now making it possible to edit the DNA in the cells of early embryos before they are placed into the woman's uterus.

(i) Name this type of gene therapy.

_____ [1]

(ii) Explain fully a possible health benefit in editing the DNA at this early stage of development.

_____

_____

_____

_____ [2]

(iii) Suggest an advantage of the process taking place when undergoing in-vitro fertilisation rather than later during pregnancy.

_____

_____ [1]

(b) Blood cells are made in the bone marrow.

Leukaemia is a form of cancer caused by errors in the DNA of bone marrow cells.

One treatment method involves removing bone marrow from a patient and replacing it with healthy bone marrow cells (transplant) from a donor.

The donor needs to be a good tissue match with the patient to reduce the risk of the transplant being rejected.

However, for the transplant method to work, the treatment may need to be repeated several times.

Gene therapy can now be used to treat some forms of leukaemia.

This involves editing the DNA in the bone marrow cells of the person with leukaemia.

(i) Using the information provided and your knowledge, suggest three benefits of using gene editing rather than transplants from a donor to treat leukaemia.

1. _____

_____

2. _____

_____

3. _____

_____ [3]

The form of gene therapy described above involves repairing defective genes.

(ii) Apart from **repairing** defective genes, state **one** other method of gene therapy.

_____

_____ [1]

[Turn over

9 **(a)** Define the term **recessive allele**.

_____

_____

_____ [1]

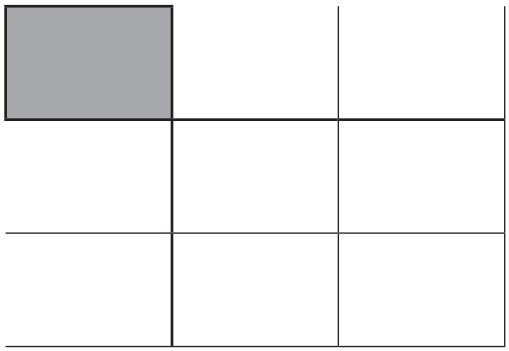
(b) Batten disease is a rare, life-limiting, genetic human condition.

The allele for Batten disease is **not** sex-linked and is recessive.

Using a Punnett Square, **show** how it is possible for two parents, neither of whom have Batten disease, to have a child who has the condition.

In your answer, use **B** for the dominant allele and **b** for the recessive allele.

Put a circle around the individual(s) with Batten disease.



[4]

There are four blood groups in the human ABO system. Individuals can have blood groups A, B, AB or O.

There are three alleles (I^A, I^B and I^O) that determine the blood groups of individuals. The table below shows the four blood groups and their possible genotypes.

Blood group (phenotype)	Possible genotypes
A	I^AI^A and I^AI^O
B	I^BI^B and I^BI^O
AB	I^AI^B
O	I^OI^O

(c) Using the table, state the evidence which shows that the alleles I^A and I^B are co-dominant.

_____

_____

_____ [1]

[Turn over

(d) (i) A person who does **not** have Batten disease and is blood group O can have two possible genotypes.
One of these is **BB I^OI^O**.
Give the other possible genotype.

_____ [1]

A man with genotype **Bb I^AI^O** had children with a woman with the genotype **Bb I^OI^O**.

(ii) Give the four possible types of gametes the man with the genotype **Bb I^AI^O** could produce.

_____ _____

_____ _____ [2]

(iii) Give the two possible types of gametes the woman with the genotype **Bb I^OI^O** would produce.

_____ and _____ [1]

(iv) Complete the Punnett Square below to show the parental gametes and the genotypes of the offspring.

[3]

(v) Give the numbers of each of the phenotypes produced in this Punnett Square.

Does not have Batten disease and is blood group A _____

Does not have Batten disease and is blood group O _____

Has Batten disease and is blood group A _____

Has Batten disease and is blood group O _____ [2]

(vi) Give the probability of these parents having a child who has Batten disease and is also blood group O.

_____ [1]

[Turn over

(e) A species of beetle can have green or black wing cases and long or short wings.

The alleles for green wing cases and long wings are dominant.

In a cross between two beetles, each heterozygous for wing case colour and wing length, the following offspring numbers were obtained.

green wing cases and long wings: 93

green wing cases and short wings: 27

black wing cases and long wings: 33

black wing cases and short wings: 7

A chi-square test was carried out to check if the offspring numbers matched the expected 9 : 3 : 3 : 1 ratio.

A calculated χ^2 value of 1.6 was obtained.

The table below gives probability values for use in a χ^2 test.

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

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

_____ [1]

(ii) Between which range of probabilities does the χ^2 fit?

_____ and _____ [1]

(iii) State the outcome of the statistical test.

_____

_____

_____ [1]

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