# Genome Projects and Recombinant DNA: AQA A Level Biology Exam Qu

18 questions · 40 marks · about 52 minutes. Answer in the spaces, then check the mark schemes at the end.

**Question 1 · information from earlier in the question**

0 3 In Europe, viruses have infected a large number of frogs of different species. The viruses are closely related and all belong to the Ranavirus group.

Previously, the viruses infected only one species of frog.

**Question 1 · A level Paper 3 · November 2020 · Q03.3 · 2 marks**

0 3 . 3 Determining the genome of the viruses could allow scientists to develop a vaccine.

Explain how.

**[2 marks]**

**Question 2 · A level Paper 2 · June 2025 · Q08.2 · 2 marks**

The genome of the bacterium *Escherichia coli* is approximately 4.6 million base pairs in length and contains around 4000 genes.

0 8 . 2 The analysis of the genome of *E. coli* allows the proteome to be determined more easily than that of eukaryotes.

Explain why.

**[2 marks]**


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**Question 3 · information from earlier in the question**

**0 3** In Europe, viruses have infected a large number of frogs of different species. The viruses are closely related and all belong to the Ranavirus group.

Previously, the viruses infected only one species of frog.

**Question 3 · A level Paper 3 · November 2020 · Q03.2 · 1 mark**

**0 3 . 2** Name **two** techniques the scientists may have used when analysing viral DNA to determine that the viruses were closely related.

**[1 mark]**

1 _____

2 _____

**Question 4 · information from earlier in the question**

**1 0** Read the following passage.

The wall gecko is a medium-sized lizard. In an isolated habitat of southern Italy, the wall gecko shows phenotypic diversity. Scientists investigated whether disruptive selection was leading to sympatric speciation in the wall gecko.

Pale geckos live only on walls and are nocturnal (active at night). Dark geckos live mainly on the dark trunks of olive trees and are diurnal (active during the day). These diurnal geckos can change skin colour when occupying different surfaces during the day.

Comparison of mitochondrial genes indicated that the diurnal geckos formed a distinct genetic group. This comparison also confirmed that all the geckos in the habitat were of the same species.

The scientists used the mark-release-recapture method to estimate the size of the population of geckos in the habitat.

Use the information in the passage and your own knowledge to answer the following questions.


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**Question 4 · A level Paper 2 · June 2024 · Q10.4 · 3 marks**

**1 0 . 4** Explain how comparison of mitochondrial genes could indicate that the nocturnal geckos formed a distinct genetic group (lines 9–10).

In your answer, explain how new techniques enable the comparison of genes to be completed rapidly.

**[3 marks]**


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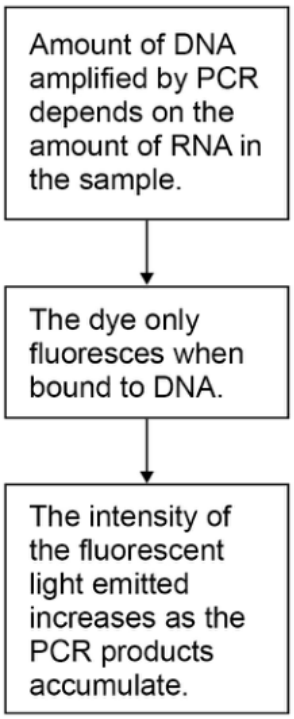
**0 8** One way to detect and measure accurately the amount of RNA in a tissue sample is by RT-PCR (reverse transcriptase-polymerase chain reaction).

RT-PCR uses a reaction mixture containing:

- the sample for testing
- reverse transcriptase
- DNA nucleotides
- primers
- DNA polymerase
- fluorescent dye.

The principle behind this method is shown in **Figure 8**.

**Figure 8**




**0 8 . 1** Explain the role of reverse transcriptase in RT-PCR.

**[1 mark]**


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**Question 6 · A level Paper 2 · June 2023 · Q09.1 · 2 marks**

0 9 Short tandem repeats (STRs) are short sequences of DNA, usually 2 to 7 base pairs. STRs are repeated a number of times, one after another. For example, the STR D5S818 is made up of AGAT repeated 7 to 16 times.

STRs are found throughout the whole genome.

The repeated sequences in STRs are common to all humans. Due to variation in the number of repeats, STRs can be used in genetic fingerprinting.

0 9 . 1 Describe how STRs could be removed from a sample of DNA.

**[2 marks]**

**Question 7 · A level Paper 2 · November 2021 · Q08.1 · 4 marks**

0 8 . 1 Describe and explain how the polymerase chain reaction (PCR) is used to amplify a DNA fragment.

**[4 marks]**


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**Question 8 · information from earlier in the question**

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STRs are found throughout the whole genome.

The repeated sequences in STRs are common to all humans. Due to variation in the number of repeats, STRs can be used in genetic fingerprinting.

**Question 8 · A level Paper 2 · June 2023 · Q09.2 · 2 marks**

**0 9 . 2** Genetic fingerprinting using STRs requires amplification of the STRs using the polymerase chain reaction (PCR). The short base sequences either side of a specific STR are known.

Explain the importance of knowing these base sequences in PCR.

**[2 marks]**


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**Question 9 · information from earlier in the question**

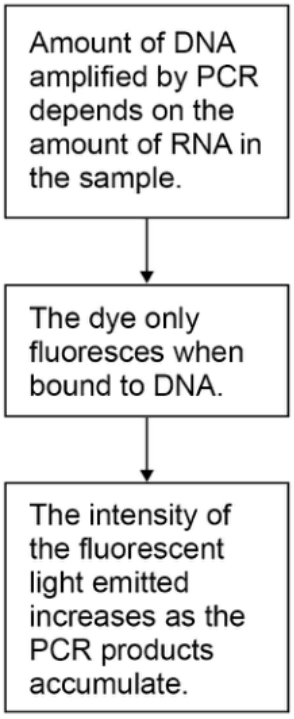
**0 8** One way to detect and measure accurately the amount of RNA in a tissue sample is by RT-PCR (reverse transcriptase-polymerase chain reaction).

RT-PCR uses a reaction mixture containing:

- the sample for testing
- reverse transcriptase
- DNA nucleotides
- primers
- DNA polymerase
- fluorescent dye.

The principle behind this method is shown in **Figure 8**.

**Figure 8**





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**Question 9 · A level Paper 2 · June 2017 · Q08.3 · 2 marks**

**0 8 . 3** Any DNA in the sample is hydrolysed by enzymes before the sample is added to the reaction mixture.

Explain why.

**[2 marks]**

**Question 10 · A level Paper 2 · June 2017 · Q08.6 · 2 marks**

**0 8 . 6** Scientists have used the RT-PCR method to detect the presence of different RNA viruses in patients suffering from respiratory diseases.

The scientists produced a variety of primers for this procedure.

Explain why.

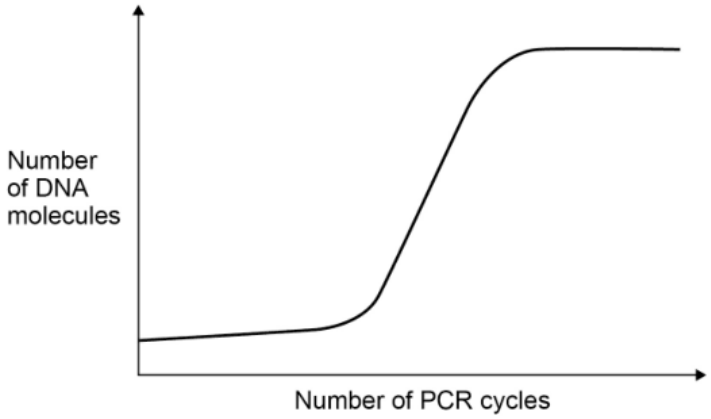
**[2 marks]**


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**Question 11 · A level Paper 2 · November 2021 · Q08.2 · 2 marks**

**Figure 9** shows the number of DNA molecules produced using a PCR.

**Figure 9**




**0 8 . 2** Explain the shape of the curve in **Figure 9**.

**[2 marks]**

**Question 12 · information from earlier in the question**

**0 9** Short tandem repeats (STRs) are short sequences of DNA, usually 2 to 7 base pairs. STRs are repeated a number of times, one after another. For example, the STR D5S818 is made up of AGAT repeated 7 to 16 times.

STRs are found throughout the whole genome.

The repeated sequences in STRs are common to all humans. Due to variation in the number of repeats, STRs can be used in genetic fingerprinting.


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**Question 12 · A level Paper 2 · June 2023 · Q09.3 · 2 marks**

**0 9 . 3** A single STR molecule consisting of a 12 **base pair** sequence of AGAT was amplified 50 times using PCR.

Calculate the total number of base pairs in all the STR molecules after 50 cycles of PCR.

Show your working.

**[2 marks]**

Answer _____

**Question 13 · A level Paper 2 · June 2024 · Q07.4 · 2 marks**

**0 7 . 4** Describe how restriction endonuclease and DNA ligase are used to insert a gene into a plasmid.

**[2 marks]**

**Question 14 · information from earlier in the question**

Recombinant DNA technology can involve the transfer of fragments of human DNA into bacteria. The bacteria are then used to produce human proteins.


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**Question 14 · A level Paper 3 · June 2022 · Q04.4 · 2 marks**

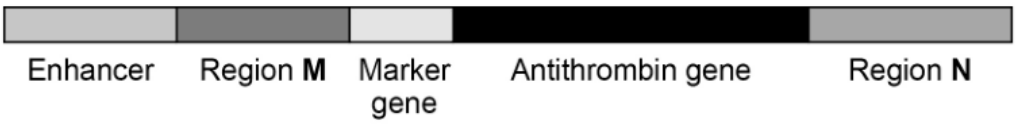
Antithrombin is a protein. Antithrombin prevents blood from clotting too much.

Some people have a deficiency of antithrombin in their blood, so they need to inject the protein.

Genetically modified goats are used to produce this protein. The human antithrombin gene is transferred into goat embryos. The adult goats then make human antithrombin protein.

**Figure 7** shows an example of a DNA fragment that can be transferred into the cells of goats.

**Figure 7**




**0 4 . 4** The enhancer stimulates region **M**.

Name regions **M** and **N** shown in **Figure 7**.

**[2 marks]**

Region **M** _____

Region **N** _____

**Question 15 · A level Paper 3 · June 2022 · Q04.5 · 1 mark**

**0 4 . 5** Explain the purpose of the marker gene.

**[1 mark]**

_____

_____

**Question 16 · information from earlier in the question**

**0 9** A scientist produced transgenic zebrafish.

She obtained a gene from silverside fish. The gene codes for a growth hormone (GH).

She inserted copies of this *GH* gene into plasmids. She then microinjected these recombinant plasmids into fertilised egg cells of zebrafish.


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**Question 16 · A level Paper 2 · November 2020 · Q09.2 · 2 marks**

**0 9 . 2** Microinjection of DNA into fertilised egg cells is a frequent method of producing transgenic fish. However, the insertion of the transferred gene into nuclear DNA may be delayed. Consequently, the offspring of transgenic fish may not possess the desired characteristic.

Suggest and explain how delayed insertion of the *GH* gene could produce offspring of transgenic fish without the desired characteristic.

**[2 marks]**

**Question 17 · information from earlier in the question**

**0 2** Sickle cell disease (SCD) is a group of inherited disorders. People with SCD have sickle-shaped red blood cells. A single base substitution mutation can cause one type of SCD. This mutation causes a change in the structure of the beta polypeptide chains in haemoglobin.

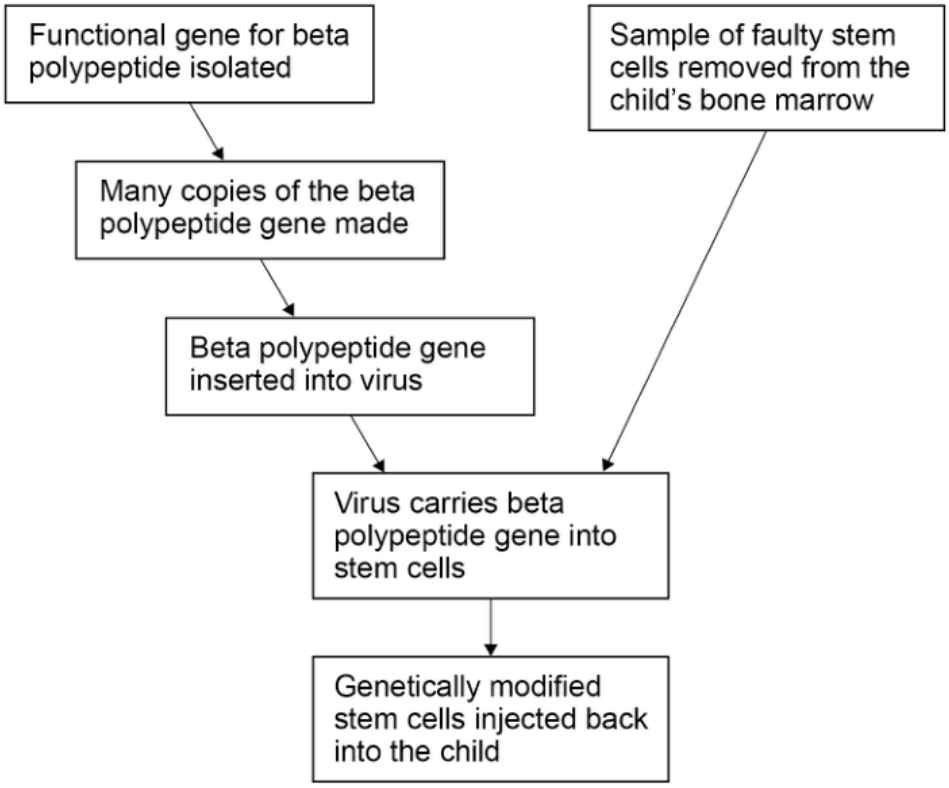
Haematopoietic stem cell transplantation (HSCT) is a long-term treatment for SCD. In HSCT, the patient receives stem cells from the bone marrow of a person who does not have SCD. The donor is often the patient's brother or sister. Before the treatment starts, the patient's faulty bone marrow cells have to be destroyed.


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A new long-term treatment for SCD involves the use of gene therapy.

**Figure 2** shows some of the stages involved in this treatment in a child with SCD.

**Figure 2**




**0 2 . 3** Some scientists have concluded that this method of gene therapy will be a more effective long-term treatment for SCD than HSCT. Use all the information provided to evaluate this conclusion.

**[3 marks]**


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**Question 18 · information from earlier in the question**

**0 4 . 1** People with the inherited blood disorder haemophilia A (HA) have a reduced ability to clot blood. One variant of HA is due to an inversion mutation in the *F8* gene. This mutation causes the production of a non-functional form of the protein, factor VIII. Factor VIII is **not** an enzyme.

Explain how an inversion mutation can cause the production of non-functional factor VIII.

**[4 marks]**


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**0 4 . 2** Currently, the most effective treatment for HA is weekly or monthly injections of factor VIII. Scientists investigated the effectiveness of a gene therapy product, Roctavian, as a treatment for HA. Roctavian contains a viral vector that has the DNA coding for factor VIII.

The scientists:

- selected 112 male volunteers with severe HA, who were at least 18 years of age
- treated each volunteer with a single injection of Roctavian into their blood
- recorded the number of bleeding episodes (internal bleeding) in these volunteers over a two-year period (set **A** results)
- compared these data with the number of bleeding episodes per year in the volunteers **before** receiving Roctavian (set **B** results).

**Table 1** shows the scientists' results.

A value of ±2 × SD from the mean includes over 95% of the data.

**Table 1**

| Volunteer set | Mean number of bleeding episodes per year (±2 × SD) |
|---|---|
| A (Received Roctavian) | 0.8 ± (1.2) |
| B (Received factor VIII) | 4.8 ± (0.6) |

Use all the information provided to evaluate the use of Roctavian as a treatment for HA.

**[5 marks]**


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## Mark schemes

**Question 1 · A level Paper 3 · November 2020 · Q03.3 · 2 marks**

| | | | |
|---|---|---|---|
| 03.3 | 1. (The scientists) could identify proteins (that derive from the genetic code)<br><br>**OR**<br><br>(The scientists) could identify the proteome;<br><br>2. (They) could (then) identify potential antigens (to use in the vaccine); | 2 | 2. Reject if answer suggests vaccine contains antibodies |

**Question 2 · A level Paper 2 · June 2025 · Q08.2 · 2 marks**

| | |
|---|---|
| 08.2 | 1. (*E. coli* / prokaryotes/bacteria) do not have introns /non-coding (DNA)<br><br>**OR**<br><br>*(E. coli* / prokaryotes/bacteria) only have exons / coding (DNA)<br><br>**OR**<br><br>Eukaryotes contain introns / non-coding (DNA);<br><br>2. Eukaryotes contain (more) regulatory genes; |

| | |
|---|---|
| 2 (2 x AO1) | 1 and 2 Accept 'It' refers to *E. coli*<br><br>2. Accept prokaryotes have no/fewer regulatory genes |


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**Question 3 · A level Paper 3 · November 2020 · Q03.2 · 1 mark**

| | | | |
|---|---|---|---|
| **03.2** | For **one** mark, accept any **two** of the following:<br>• The polymerase chain reaction<br>• Genetic/DNA fingerprinting<br>• (Gel) electrophoresis<br>• DNA/genome sequencing; | 1 | Accept PCR for polymerase chain reaction<br>Accept autoradiography<br>Accept DNA hybridisation<br>Accept compare DNA/base sequence for 'DNA sequencing'<br>Ignore compare mRNA base sequence<br>Ignore compare amino acid sequence<br>Ignore DNA probes |


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| | | |
|---|---|---|
| **10.4** | 1. Compare <u>DNA</u> base/nucleotide sequence<br>**OR**<br>Compare banding/position of DNA fragments;<br>2. A distinct (group) will have different alleles/DNA/banding (from other group/s)<br>**OR**<br>If **not** distinct (group) will have similar alleles/DNA/banding (to other group/s)<br>3. DNA sequencing is automated/computerised<br>**OR**<br>Genetic/DNA fingerprinting is automated/ computerised<br>**OR**<br>PCR amplifies DNA/genes<br>**OR**<br>Genetic fingerprinting/electrophoresis separates fragments/genes/alleles<br>**OR**<br>Use of DNA probes/hybridisation to identify genes/alleles; | 3<br>(3 x AO2) |

1. Idea of 'comparison' must be conveyed.

1. Accept alleles/VNTRs for 'DNA fragments'.

1 and 2. Accept genes for 'DNA fragments' in 1 but reject genes in mark point 2.


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**Question 5 · A level Paper 2 · June 2017 · Q08.1 · 1 mark**

| | | | |
|---|---|---|---|
| 08.1 | Produces (c)DNA using (m)RNA; | 1 | Accept: 'converts' (m)RNA to (c)DNA.<br>Reject: tRNA |

**Question 6 · A level Paper 2 · June 2023 · Q09.1 · 2 marks**

| | | | |
|---|---|---|---|
| **09.1** | 1. Restriction endonucleases/enzymes;<br>2. (Cut DNA) at specific base sequences/pairs<br>**OR**<br>(Cut DNA) at recognition/restriction sites; | 2<br>(2 x AO2) | 2. Accept 'at palindromic sequences' |

**Question 7 · A level Paper 2 · November 2021 · Q08.1 · 4 marks**

| | | | |
|---|---|---|---|
| **08.1** | 1. (Requires DNA fragment) DNA polymerase, (DNA) nucleotides and primers;<br>2. Heat to 95 °C to break hydrogen bonds (and separate strands);<br>3. Reduce temperature so primers bind to DNA/strands;<br>4. Increase temperature, DNA polymerase joins nucleotides (and repeat method); | 4 | 1 and 4. Accept Taq polymerase for DNA polymerase.<br>2. Accept temperature in range 90 to 95 °C.<br>3. Accept temperature in range 40 to 65 °C.<br>4. Accept temperature in range 70 to 75 °C. |

**Question 8 · A level Paper 2 · June 2023 · Q09.2 · 2 marks**

| | | |
|---|---|---|
| **09.2** | 1. (For) primers;<br>2. (To produce) a complementary base sequence<br>**OR**<br>(Primers provide starting sequence) for DNA/taq polymerase<br>**OR**<br>(Primers) stop (original) DNA strands re-joining; | 2<br>(2 x AO2) |


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**Question 9 · A level Paper 2 · June 2017 · Q08.3 · 2 marks**

| | | | |
|---|---|---|---|
| 08.3 | 1. To remove any DNA present;<br>2. As this DNA would be amplified/replicated; | 2 | 1. Must be idea of removal/destruction.<br>2. Accept: idea of DNA not being used as template. |

**Question 10 · A level Paper 2 · June 2017 · Q08.6 · 2 marks**

| | | | |
|---|---|---|---|
| 08.6 | 1. Base sequences differ;<br>2. (Different) <u>complementary</u> primers required; | 2 | 1. Accept: reference to either RNA or DNA base sequences but reject reference to DNA base sequence in viruses. |

**Question 11 · A level Paper 2 · November 2021 · Q08.2 · 2 marks**

| | | | |
|---|---|---|---|
| **08.2** | 1. (Initially) number (of molecules) doubling is low<br>**OR**<br>Doubles each cycle to produce exponential increase;<br>2. Plateaus as no more nucleotides/primers; | 2 | 1. First alternative relates to idea of low numbers i.e., 2, 4, 8, 16, 32 etc.<br>2. Accept 'levels out' or 'flattens' for plateaus.<br>2. Accept enzyme/polymerase (eventually) denatures. |


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| | | |
|---|---|---|
| **09.3** | Correct answer of $1.35 \times 10^{16}$ = **2 marks**<br>**OR**<br>Correct answer of $1.36 \times 10^{16}$ (due to rounding at an earlier stage of the calculation) = **2 marks**<br>**OR**<br>Correct answer of $1.4 \times 10^{16}$ = **2 marks**;;<br>Incorrect answer but shows $2^{50}$ = **1 mark**; | 2<br>(2 x AO2) |

Ignore any numbers after 1.35.


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**Question 13 · A level Paper 2 · June 2024 · Q07.4 · 2 marks**

| | | |
|---|---|---|
| **07.4** | 1. Restriction endonucleases/enzymes cuts <u>plasmid</u><br>**OR**<br>Restriction endonucleases/enzymes produce 'sticky ends';<br>2. Ligase joins gene/DNA and plasmid<br>**OR**<br>Ligase joins 'sticky ends'<br>**OR**<br>Ligase forms phosphodiester bonds; | 2<br>(2 x AO1) |

1. Accept 'cuts DNA of <u>plasmid</u>'.

1. Ignore restriction enzymes cuts out the gene.

2. Ignore reference to hydrogen bonds or joining complementary base pairs.

**Question 14 · A level Paper 3 · June 2022 · Q04.4 · 2 marks**

| | | | |
|---|---|---|---|
| **04.4** | 1. (Region **M**) promoter;<br>2. (Region **N**) terminator; | 2<br>(2 x AO2) | Accept phonetic spellings |


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**Question 15 · A level Paper 3 · June 2022 · Q04.5 · 1 mark**

| | | | |
|---|---|---|---|
| 04.5 | Shows that the (antithrombin) gene has been taken up (by cells/embryos/goats)<br>**OR**<br>Shows transgenic/transformed goat cells/goat embryos/goats<br>**OR**<br>Allows detection of genetically modified cells/ organisms/mammals/goats; | 1 (AO1) | Accept 'GM' for 'genetically modified' |

**Question 16 · A level Paper 2 · November 2020 · Q09.2 · 2 marks**

| | | | |
|---|---|---|---|
| 09.2 | 1. Cell division has occurred (before gene added);<br>2. (Cells producing) gametes do not receive the gene; | 2 | 1. Accept mitosis but reject meiosis.<br>1. Accept DNA replication has occurred. |


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| | |
|---|---|
| 02.3 | (For gene therapy)<br>1. No destruction of bone marrow<br>**OR**<br>No destruction of stem cells;<br>2. Donors are not required;<br>3. Less/no chance of rejection (own stem cells);<br>(Against gene therapy)<br>4. Sickle/faulty (red blood) cells still produced<br>5. Immune response against genetically modified cells/virus<br>**OR**<br>Long-term effect not known (as is new treatment)<br>**OR**<br>Virus could cause side effects; |

| | |
|---|---|
| 3 max | **Max 2 marks** for marking points 1, 2 and 3<br>1. Accept no destruction of faulty bone marrow unless context indicates this is against gene therapy.<br>2. Stating 'only own cells used' is **not** equivalent.<br>5. Accept 'virus could cause problems' or |


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| | | |
|---|---|---|
| 04.2 | **Mark point 1 required for maximum marks.**<br>**Max 3 marks from mark points 4 to 7**<br>**(For)**<br>1. SDs do not overlap so significant difference/decrease/change (with Roctavian);<br>2. Only one injection/treatment is required<br>**OR**<br>Weekly/monthly/regular treatments/injections not required;<br>3. (Investigation) compares same volunteers;<br>**(Against)**<br>4. Only males (investigated/treated)<br>OR<br>No females (investigated/treated);<br>5. (Possible) side effects;<br>6. At least 18 (years of age investigated/treated)<br>**OR**<br>No under 18 (years of age investigated/treated);<br>7. Only severe HA (investigated/treated); | 5 max (5 x AO3) |

1. Reject 'results are significant'

1. Accept 'not due to chance' for 'significant'

2. Accept fewer/less for 'one'

Ignore 'sample size'

'length of investigation' and


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