Regulation of Transcription and Translation: AQA A Level Biology Exam

18 questions · 44 marks · about 57 minutes. Answer in the spaces, then check the mark schemes at the end.

Question 1 · information from earlier in the question

0 2 Testosterone is a steroid hormone that belongs to a group of male sex hormones called androgens.

Question 1 · A level Paper 2 · November 2020 · Q02.3 · 2 marks

0 2 . 3 The binding of testosterone to an AR changes the shape of the AR. This AR molecule now enters the nucleus and stimulates gene expression.

Suggest how the AR could stimulate gene expression.

[2 marks]

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0 6 . 3 Scientists investigated the use of induced pluripotent stem cells (iPS cells) to treat type I diabetes in mice. The scientists used four transcription factors to reprogramme skin cells to form iPS cells. The scientists then stimulated the *in vitro* differentiation of iPS cells into pancreatic cells.

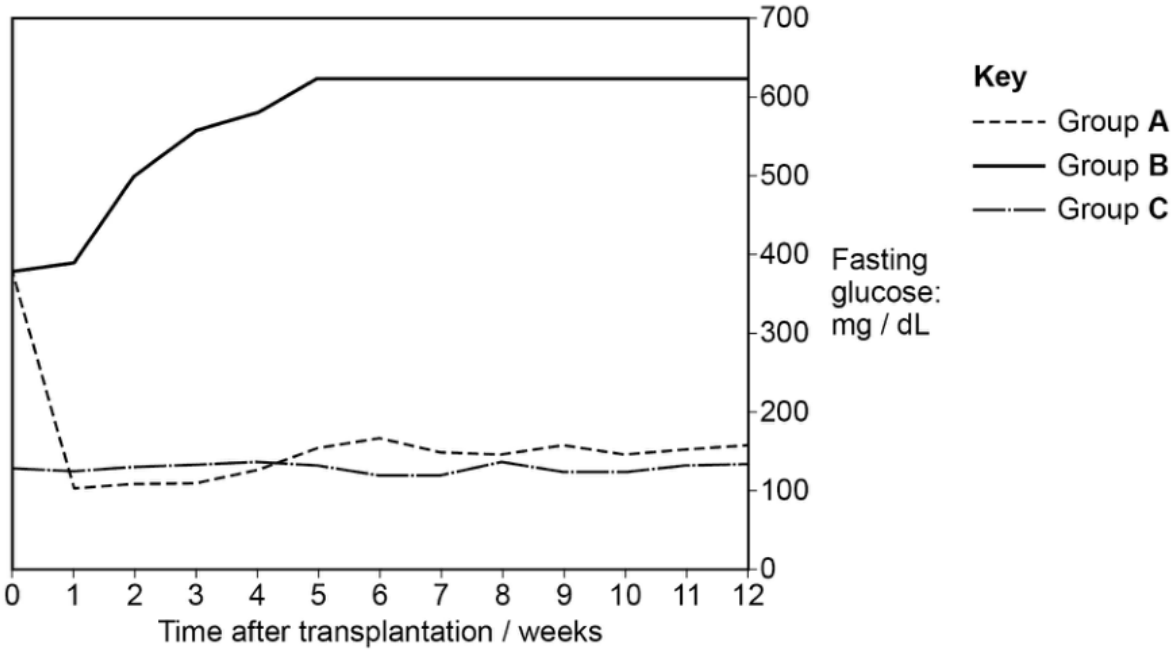
The scientists set up three experimental groups:

- Group **A** – 30 mice with type I diabetes received pancreatic cell transplants derived from iPS cells.
- Group **B** – 30 mice with type I diabetes were left untreated.
- Group **C** – 30 mice without diabetes were left untreated.

The scientists measured the blood glucose concentration of all the mice on a weekly basis for 12 weeks.

The results the scientists obtained are shown in **Figure 6**.

Figure 6



Suggest how transcription factors can **reprogramme** cells to form iPS cells.

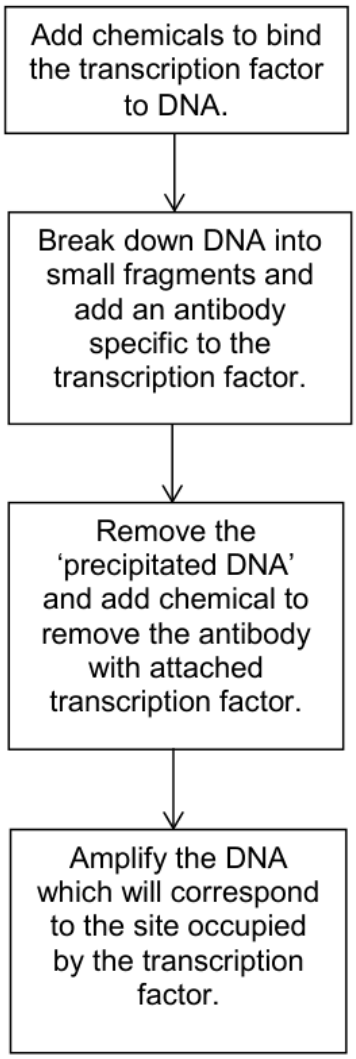
[2 marks]

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Question 3 · information from earlier in the question

Chromatin immunoprecipitation is one method to determine where a transcription factor binds to DNA. The principle behind this procedure is shown in **Figure 8**.

Figure 8



Question 3 · A level Paper 2 · June 2018 · Q08.3 · 1 mark

0 8 . 3 Use **Figure 8** to explain what 'precipitated DNA' consists of.

[1 mark]

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Question 4 · information from earlier in the question

0 9 Lactose is the main sugar in milk and is hydrolysed by the enzyme lactase. Lactase is essential to newborn mammals as milk is their only source of food. Most mammals stop producing lactase when they start feeding on other food sources. Humans are an exception to this because some continue to produce lactase as adults. The ability to continue producing lactase is known as lactase persistence (LP) and is controlled by a dominant allele. A number of hypotheses based on different selection pressures have been put forward to explain LP in humans.

Question 4 · A level Paper 2 · June 2018 · Q09.3 · 2 marks

0 9 . 3 Lactase persistence is caused by a mutation in DNA. This mutation does **not** occur in the gene coding for lactase.

Suggest and explain how this mutation causes LP.

[2 marks]

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Question 5 · information from earlier in the question

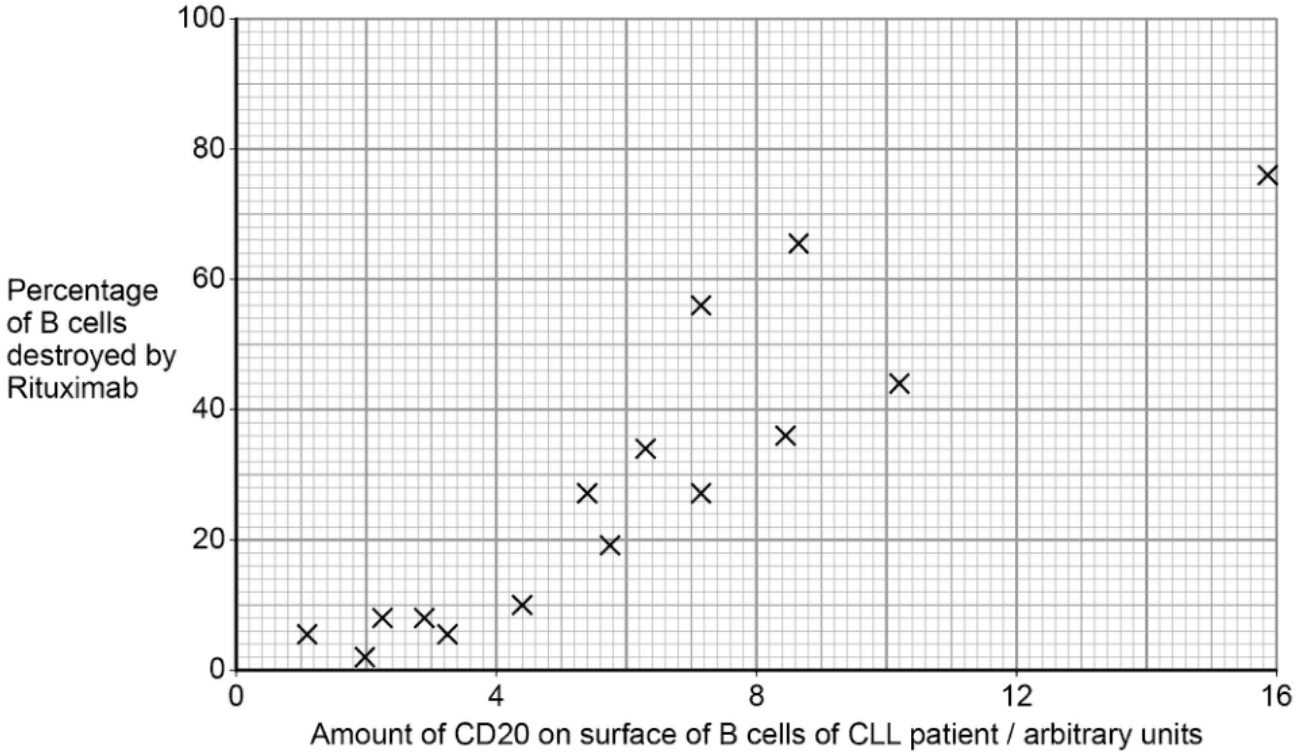
0 2 Chronic lymphocytic leukaemia (CLL) is a cancer that affects some B cells of a person's immune system.

Rituximab is a drug used to treat CLL. It binds to a protein called CD20 on the surface of B cells. If enough Rituximab binds to a B cell, it can kill the cell. Rituximab kills **both** healthy **and** cancerous B cells. The body then produces new B cells.

The amount of CD20 on the surface of B cells varies from one person to another. Doctors investigated the relationship between the amount of CD20 on the B cells of a patient and the percentage of B cells destroyed by Rituximab.

Figure 2 shows the doctors' results. Each cross is the result for one patient.

Figure 2



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Approximately 11 percent of people with CLL also have mutations of a gene called *NOTCH1*. This leads to production of a non-functional transcription factor associated with CD20 production.

The doctors determined the median percentage of B cells destroyed by Rituximab in people with CLL who had the *NOTCH1* mutation and those who did not.

The doctors' results are shown in **Table 1**.

Table 1

	Median percentage of B cell destroyed by Rituximab
In people with CLL who had the *NOTCH1* mutation	**4**
In people with CLL who did **not** have *NOTCH1* mutation	**22**

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Question 5 · A level Paper 3 · June 2018 · Q02.4 · 3 marks

0 2 . 4 Use all of the information to suggest how the mutation of *NOTCH1* led to the difference in the percentage of B cells destroyed by Rituximab.

[3 marks]

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0 5 Cleft lip and palate (CLP) is a split in the upper lip and/or the roof of the mouth. CLP is present from birth.

CLP can be caused by various genetic and environmental factors.

CDH1 is a gene involved in lip and mouth development in a fetus before a baby is born.

Scientists hypothesised that CLP is caused by a 'two-hit' model:

- a 'genetic hit' through loss of *CDH1* function
- an 'environmental hit' leading to *CDH1* methylation caused when inflammation affects a fetus.

Inflammation can result from the mother having an infection or diabetes.

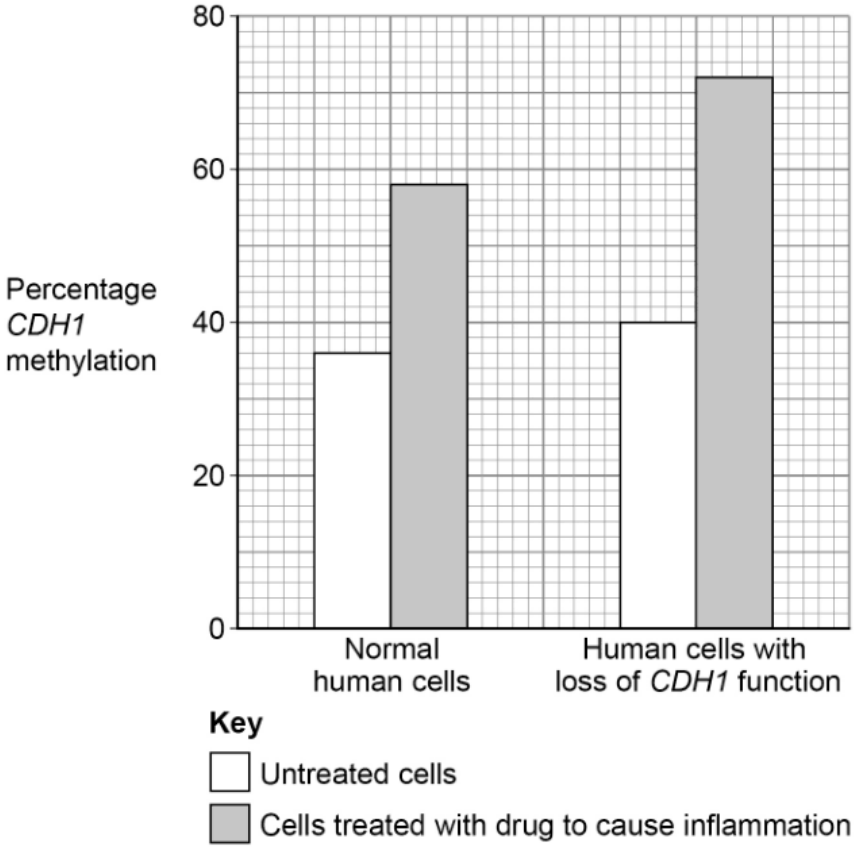
Scientists investigated the 'two-hit' model using human cells grown *in vitro*.

They:

- grew normal cells and cells with a loss of *CDH1* function
- treated some of each type of cell with a drug to cause inflammation and left some of each type of cell untreated
- measured the percentage *CDH1* methylation of the cells.

Figure 4 shows the scientists' results.

Figure 4



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Question 6 · A level Paper 3 · June 2025 · Q05.2 · 1 mark

0 5 . 2 Describe **one** effect methylation could have on *CDH1*

[1 mark]

Question 7 · information from earlier in the question

1 0 Alzheimer's disease (AD) is a non-reversible brain disorder that develops over a number of years. At the start of 2014 the number of Americans with AD was estimated to be 5.4 million. Every 30 seconds another person in America develops AD.

In the brain of a person with AD there is a lower concentration of acetylcholine. This affects communication between nerve cells and initially results in memory loss and confusion. Some of the symptoms of AD that are associated with communication between nerve cells are reduced by taking the drug donepezil. Donepezil inhibits the enzyme acetylcholinesterase.

A gene mutation called E280A found on chromosome 14 causes early-onset AD at a mean age of 49 years. The age at which the E280A mutation is expressed to cause AD varies.

Yaramul is a town in a historically isolated region of the Andes Mountains. The population of this town has the highest frequency of the E280A mutation in the world. The origin of the E280A mutation in this population has been traced back to a common ancestor in the 17th century. Natural selection has not reduced the frequency of the E280A mutation in the population.

This autosomal dominant mutation involves a change in triplet 280 from GAA to GCA. Scientists analysed chromosome 14 from 102 individuals from Yaramul. They recorded a sample size of 204 and detected 75 E280A mutations but only 74 potential AD cases. The scientists identified individuals with the mutation by whole genome sequencing. They had decided that a DNA probe would not be a suitable method to detect the E280A mutation.

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Question 7 · A level Paper 2 · June 2017 · Q10.5 · 2 marks

1 0 . 5 The age at which the E280A mutation is expressed to cause AD can vary (lines 11–12).

Suggest and explain **one** reason for this.

[2 marks]

Question 8 · information from earlier in the question

1 0 Guillain–Barré syndrome is a rare disease in which the immune system damages the myelin sheath of neurones. Myelin sheath damage can cause a range of symptoms, for example numbness, muscular weakness and muscular paralysis. Sometimes, neurones of the autonomic nervous system are affected, causing heart rate irregularities.

Huntington's disease is a disorder caused when a protein called huntingtin damages the brain. Huntingtin is produced because of a dominant, mutant allele.

The first successful drug trial to reduce concentrations of huntingtin in the human brain involved 46 patients. The patients received the drug for 4 months. The concentration of huntingtin was reduced in all the patients. The drug was injected at the base of the spine into the cerebrospinal fluid bathing the brain and spinal cord. The drug contains single-stranded DNA molecules. These single-stranded molecules inhibit the mRNA needed to produce huntingtin.

Symptoms of Huntington's disease can start at any time, but usually develop between 30 and 50 years of age. The likelihood and age when symptoms start are linked to the number of CAG base sequence repeats in the gene for Huntington's disease. However, recent studies have suggested that epigenetics may also affect the age when symptoms first start.

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Question 8 · A level Paper 2 · June 2019 · Q10.6 · 2 marks

1 0 . 6 Suggest and explain **one** way epigenetics may affect the age when symptoms of Huntington's disease start.

[2 marks]

Question 9 · information from earlier in the question

1 0 Read the following passage.

BRCA1 and *BRCA2* are human genes that code for tumour suppressor proteins. Mutations in *BRCA1* and *BRCA2* can cause cancer. Specific inherited mutations in these genes increase the risk of female breast cancers and ovarian cancers and have been associated with increased risks of several other types of cancer. Genetic testing, using DNA from saliva, can screen for all known harmful mutations in both genes.

ER-positive breast cancers have receptors for the hormone oestrogen. These cancers develop as a result of increased oestrogen concentrations in the blood. Effective treatment of ER-positive breast cancers often involves the use of drugs which have a similar structure to oestrogen.

Blood tests can be used to test for cancers. Men with prostate cancer have a high concentration of prostate-specific antigen (PSA) in their blood. Urinary infections and a naturally enlarged prostate can also increase concentrations of PSA.

Recent research has indicated that several cancers result from epigenetic abnormalities. Treatment with drugs might be able to reverse the epigenetic changes that cause cancers.

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

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Question 9 · A level Paper 2 · June 2022 · Q10.5 · 3 marks

1 0 . 5 Treatment with drugs might be able to reverse the epigenetic changes that cause cancers (lines 16–17). Suggest and explain how.

[3 marks]

Question 10 · A level Paper 2 · June 2018 · Q08.4 · 1 mark

Soybeans are used in a number of processed foods. However, soybeans contain a protein known as P34 that causes an allergic response in some people. Scientists have created transgenic soybeans that produce single-stranded cDNA, which prevents transcription of the *P34* gene. They used recombinant plasmids as vectors to transform soybean cells. After they had screened these cells for production of the P34 protein, they cultured the transformed cells to form soybean plants.

0 8 . 4 Suggest how single-stranded cDNA could prevent transcription of the *P34* gene.

[1 mark]

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Question 11 · information from earlier in the question

1 0 Guillain–Barré syndrome is a rare disease in which the immune system damages the myelin sheath of neurones. Myelin sheath damage can cause a range of symptoms, for example numbness, muscular weakness and muscular paralysis. Sometimes, neurones of the autonomic nervous system are affected, causing heart rate irregularities.

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Question 11 · A level Paper 2 · June 2019 · Q10.3 · 3 marks

1 0 . 3 The first successful drug trial to reduce concentrations of huntingtin in the brain used single-stranded DNA molecules (lines 13–14).

Suggest and explain how this drug could cause a reduction in the concentration of the protein huntingtin.

[3 marks]

Question 12 · information from earlier in the question

0 6 Plants transport sucrose from leaves to other tissues for growth and storage. SUT1 is a sucrose co-transporter protein.

Scientists investigated whether the cells of tobacco plant leaves used SUT1 to transport sucrose to other tissues.

Questions, mark schemes and examiners' report extracts © AQA.

0 6 . 2 To study the role of SUT1 in tobacco plants, scientists reduced the expression of the *SUT1* gene.

When the *SUT1* gene is transcribed, the SUT1 mRNA produced is called 'sense' SUT1 mRNA. The scientists genetically modified plants by inserting an **extra** gene so that this **also** allowed the production of 'antisense' SUT1 mRNA.

The scientists had two types of tobacco plants:

- type **A** – plants that were genetically modified
- type **B** – plants that were **not** genetically modified.

Suggest how the production of 'antisense' SUT1 mRNA in type **A** plants would reduce the expression of the *SUT1* gene.

[4 marks]

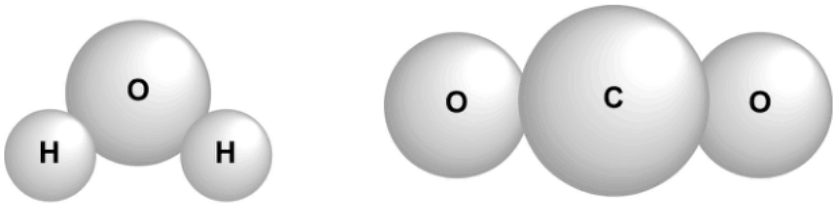
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Question 13 · information from earlier in the question

0 6 . 4 Aquaporins are channel proteins that allow the diffusion of water across membranes. One type of aquaporin, called PIP1, can also transport carbon dioxide molecules across membranes.

Figure 6 shows the structure of a water molecule and of a carbon dioxide molecule. They are drawn to the same scale.

Figure 6



Water molecule (H_2O)

Carbon dioxide molecule (CO_2)

Suggest **two** reasons why water molecules **and** carbon dioxide molecules can both pass through PIP1.

[2 marks]

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Question 13 · A level Paper 3 · June 2017 · Q06.5 · 3 marks

0 6 . 5 The scientists first produced transgenic poplar trees. These trees all had a length of foreign DNA inserted into them. This DNA led to the production of single-stranded RNA that specifically inhibited expression of the gene for PIP1.

The scientists then measured the difference in the amount of PIP1 in leaves of transgenic poplars and in leaves of wild type poplars without the foreign DNA. The amount of PIP1 in the transgenic poplars was approximately 15% of that in the wild type poplars.

Using this information, what can you conclude about the effect of the foreign DNA in the transgenic poplar trees?

[3 marks]

Question 14 · A level Paper 3 · June 2017 · Q06.6 · 1 mark

0 6 . 6 The transgenic poplars still produced some PIP1.

Suggest why.

[1 mark]

Question 15 · information from earlier in the question

0 5 Scientists investigated the role of a protein called CENP-W in mitosis. Their method involved cell fractionation and ultracentrifugation.

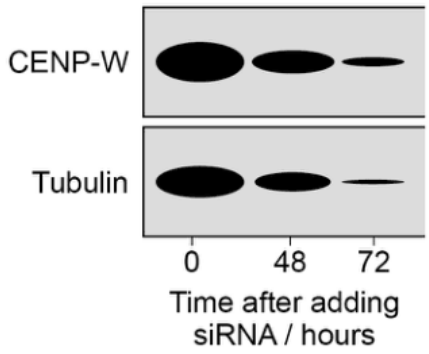
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0 5 . 3 CENP-W is involved in the formation of spindle fibres in mitosis. Spindle fibres are made of molecules of a protein called tubulin.

The scientists treated cells in a culture with small interfering RNA (siRNA). This siRNA causes RNA interference of expression of the *CENP-W* gene. The scientists took samples of cells at 0, 48 and 72 hours after adding the siRNA. They then used gel electrophoresis to separate CENP-W and tubulin from these samples.

Figure 5 shows the results of the electrophoresis. The size of each band is proportional to the amount of CENP-W or tubulin present.

Figure 5



Suggest how the siRNA produced these results. **[3 marks]**

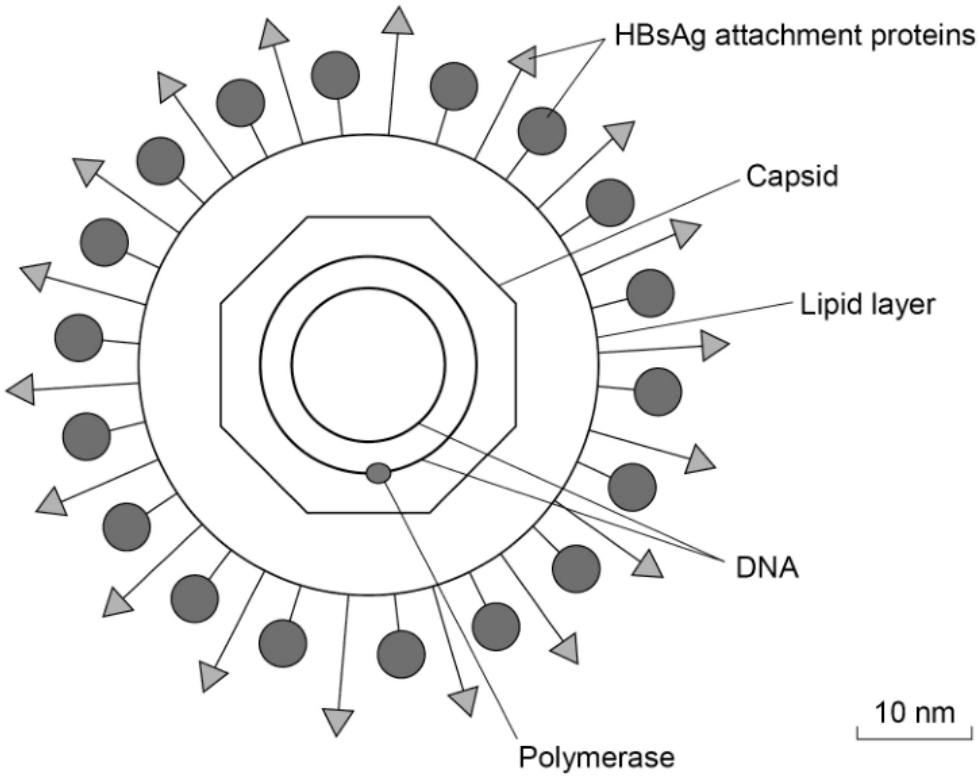
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Question 16 · information from earlier in the question

0 7 Hepatitis B is a life-threatening liver infection caused by the hepatitis B virus (HBV).

Figure 7 shows the structure of HBV.

Figure 7



Questions, mark schemes and examiners' report extracts © AQA.

Scientists investigated the effectiveness of two types of RNA interference (RNAi) molecules on reducing HBV replication. These molecules were:

- short hairpin RNA (shRNA)
- long hairpin RNA (lhRNA).

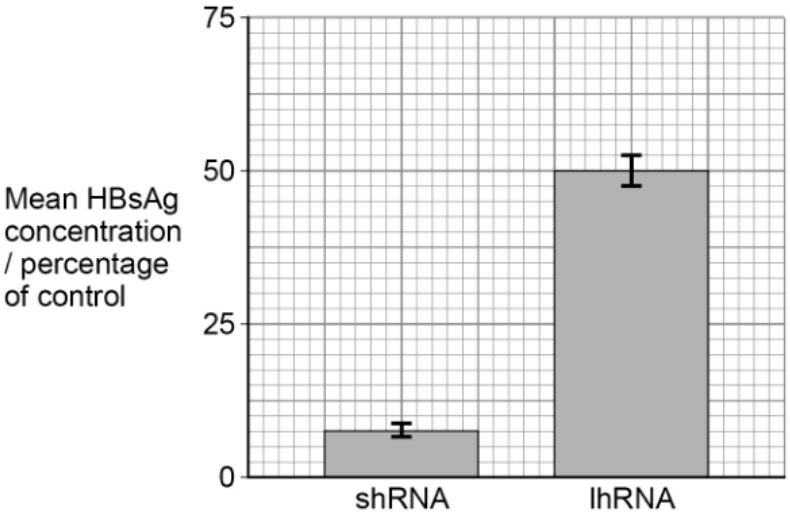
The scientists infected mouse liver cells with HBV and transferred either shRNA or lhRNA into these cells. Then they determined the concentration of the attachment proteins, HBsAg, in these cells.

The concentration of HBsAg is a measure of HBV replication.

Figure 8 shows the scientists' results.

The error bars represent ±2 standard deviations from the mean, which includes over 95% of the data.

Figure 8



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Question 16 · A level Paper 2 · November 2021 · Q07.3 · 5 marks

0 7 . 3 Using all the information provided, evaluate the use of the two types of RNAi in treating hepatitis B in humans.

Do **not** refer in your answer to how RNAi reduces HBV replication.

[5 marks]

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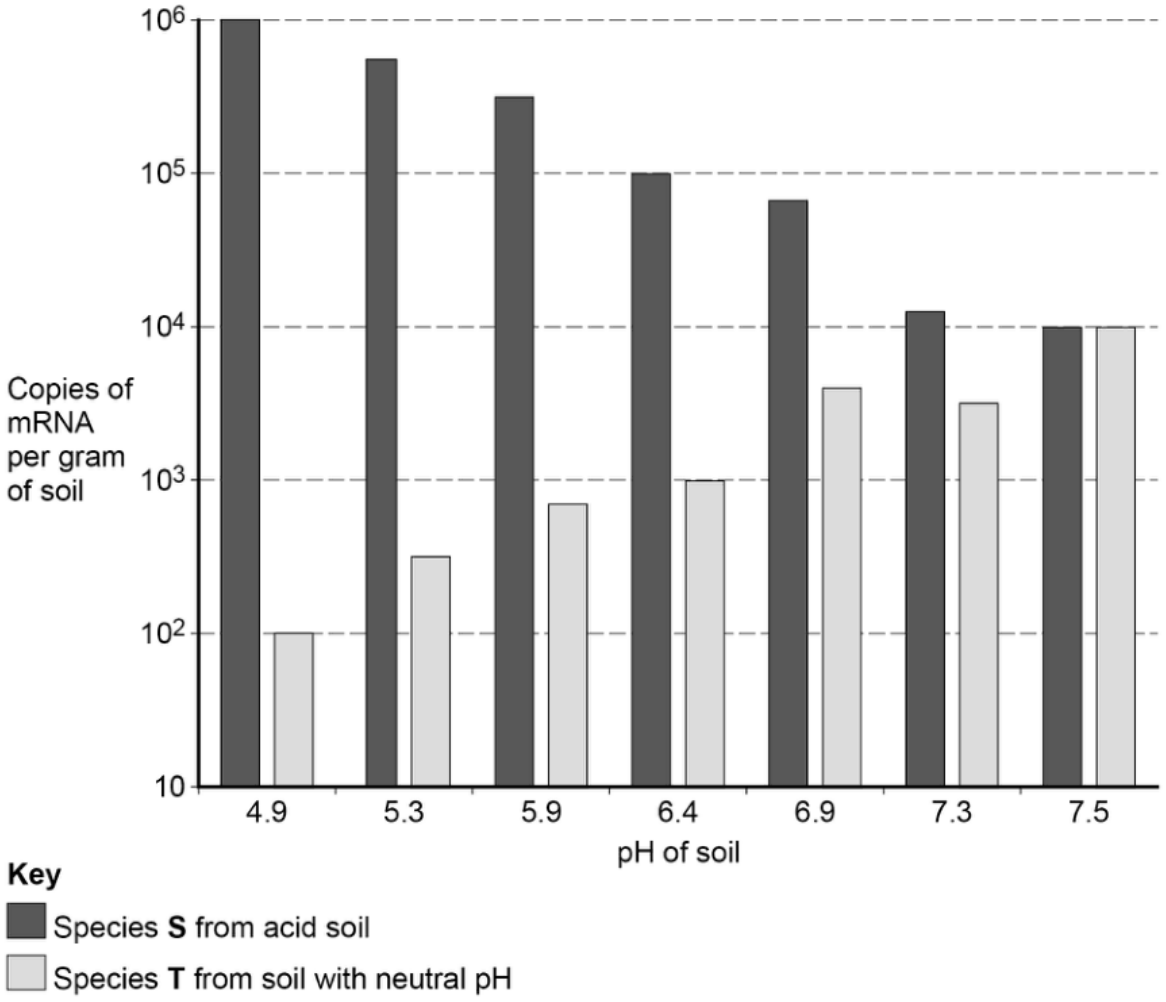
The oxidation of ammonia by nitrifying bacteria involves the enzyme ammonia monooxygenase. Each species of nitrifying bacteria has its own specific *amoA* gene that codes for production of ammonia monooxygenase.

In a second investigation, the scientists determined the expression of the *amoA* gene in two species of bacteria, **S** and **T**. Species **S** was from acid soil and species **T** was from soil with a neutral pH.

The scientists grew cultures of each species separately in soils of different pH. They determined the amount of mRNA from the *amoA* gene in each culture.

Their results are shown in **Figure 7**.

Figure 7



0 6 . 4 In which species was the number of copies of mRNA more affected by changes in soil pH from 4.9 to 7.5? Use a calculation to support your answer.

[2 marks]

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Question 18 · A level Paper 3 · June 2018 · Q06.5 · 4 marks

0 6 . 5 This method allowed the scientists to estimate the expression of the *amoA* gene in each culture but not the growth of the bacterial population in each culture.

Explain why.

[4 marks]

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Mark schemes

Question 1 · A level Paper 2 · November 2020 · Q02.3 · 2 marks

02.3	1. (AR is) a transcription factor; 2. Binds to DNA/promoter; 2 max 3. (Stimulates) RNA polymerase;	1. Ignore 'binds to bases' or 'binds to gene'. 1 and 2. Reject reference to active site, enzyme, substrate or induced fit.

Question 2 · A level Paper 2 · June 2017 · Q06.3 · 2 marks

06.3	1. Attach to gene/DNA/promoter region; 2. Stimulate/inhibit transcription/RNA polymerase;

2	Note: Genes being expressed/inhibited or switched on/off is not enough on its own.

Question 3 · A level Paper 2 · June 2018 · Q08.3 · 1 mark

08.3	1. DNA, transcription factor and antibody;	1	Accept Nucleotides for DNA Ignore 'reference to chemicals'

Question 4 · A level Paper 2 · June 2018 · Q09.3 · 2 marks

09.3	1. Mutation in promoter (DNA/gene) for transcription factor **OR** Mutation in promoter (region/DNA) for the gene **OR** Mutation in gene for transcription factor; 2. Lactase gene continues to be transcribed/active;	2	1. Accept mutation in an epistatic gene

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02.4	1. Mutation changes the tertiary structure / amino acid sequence of transcription factor; 2. Transcription factor not complementary to / cannot bind to CD20 gene / CD20 DNA; 3. Little / less / not enough / no mRNA for CD20 produced **OR** Little / less / not enough / no (mRNA for) CD20 translated / produced; 4. (Not enough CD20 so) nothing / little for Rituximab to bind to, **so** few / no B cells destroyed;	3 max

1. and 2. Do not accept unqualified statements about non-functional transcription factor – this is in stem of question

1. and 2. Answers must be in context of transcription factor, not CD20, or generic statements

2. Accept TF cannot bind to promoter (on DNA)

4. Accept converse for cells with a lot of CD20

4. Accept lower median percentage for fewer cells

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Question 6 · A level Paper 3 · June 2025 · Q05.2 · 1 mark

05.2	1. Reduce/prevent expression/transcription (of *CDH1*/the gene); 2. Prevent transcription factor binding **OR** Prevent binding to promotor (of gene); 3. Reduce/prevent production of CDH1/protein;	1 max (1 x AO1)

Reject (methylation) causes mutation

1. Accept reduce/prevent production of messenger RNA/ mRNA
1. Ignore 'switching off'
2. Accept prevent binding/activation of RNA polymerase

Question 7 · A level Paper 2 · June 2017 · Q10.5 · 2 marks

10.5	1. Epigenetics/environment/named factor e.g. stress, alcohol, toxins, diet, exercise, smoking; 2. methylation (of genes) OR acetylation (of histones);	2	1. Ignore: gender and lifestyle. 2. If further details are provided the context must be correct e.g. increased methylation or decreased acetylation inhibit gene expression/transcription.

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10.6	1. (Increased) methylation of DNA/gene/allele; 2. Inhibits/prevents transcription; **OR**	2 max
	3. Decreased methylation of DNA/gene/allele; 4. Stimulates/allows transcription; **OR** 5. Decreased acetylation of <u>histone(s)</u>; 6. Inhibits transcription; **OR** 7. Increased acetylation of <u>histone(s)</u>; 8. Stimulates/allows transcription;	

Mark in pairs but if no mark credited allow one mark for any reference to transcription or gene expression being affected.

1. Reject acetylation of DNA.

Accept gene expression for transcription but ignore gene switched on/off.

Ignore methylation of histones

Questions, mark schemes and examiners' report extracts © AQA.

Question 9 · A level Paper 2 · June 2022 · Q10.5 · 3 marks

10.5	1. (Drugs could) increase methylation of oncogene(s); 2. (Drugs could) decrease methylation of tumour suppressor gene(s); 3. (Increased) methylation of DNA/gene(s) inhibits transcription/expression (of genes) **OR** Decreased methylation of DNA/gene(s) stimulates transcription/expression (of genes); 4. Decreased acetylation of histones inhibits transcription/expression (of genes) **OR** (Increased) acetylation of histones stimulates transcription/expression (of genes);	3 max (1 x AO1, 2 x AO2)

Ignore methylation of histones and acetylation of DNA/genes.

Ignore proto-oncogenes.

3. Accept promoter (region) for DNA/gene

3 and 4. Ignore 'switching on' and 'switching off' genes once but accept as alternative(s) for 1 mark if used correctly in context of transcription/ expression for **both** points 3 and 4.

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Question 10 · A level Paper 2 · June 2018 · Q08.4 · 1 mark

08.4	1. Binds to P34 gene/DNA/mRNA **OR** Binds to transcription factor gene/DNA **OR** Binds to promoter;	1	Reject binds to transcription factor

Question 11 · A level Paper 2 · June 2019 · Q10.3 · 3 marks

10.3	1. It/DNA is <u>complementary</u> to (m)RNA; 2. Binds to mRNA (for huntingtin); 3. Prevents <u>translation</u>;	3	1. Accept (transcription) results in <u>complementary</u> (m)RNA. Ignore miRNA/siRNA/transcriptional factors. 3. Ignore transcription.

Question 12 · A level Paper 3 · November 2021 · Q06.2 · 4 marks

06.2	1. Antisense mRNA is <u>complementary</u> to 'sense' mRNA; 2. Antisense mRNA would bind/base pair to (sense) mRNA; **OR** Double stranded (m)RNA forms; 3. Ribosomes would not be able to bind; 4. Preventing/less translation (of mRNA) **OR** Preventing/less production of SUT1 (protein);	4	4. Accept descriptions of translation

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Question 13 · A level Paper 3 · June 2017 · Q06.5 · 3 marks

06.5	1. Single-stranded RNA (has base sequence) complementary to PIP1 mRNA; 2. Binds to mRNA (of PIP1)/leads to destruction of mRNA; 3. Prevents/reduces translation (of *PIP1*); 4. Reduces photosynthesis/named process that uses water;	3 max	3. Less made is insufficient

Question 14 · A level Paper 3 · June 2017 · Q06.6 · 1 mark

06.6	Not all of mRNA bound to single-stranded RNA/there is more mRNA than interfering RNA OR Not all mRNA destroyed/disabled;	1	Accept mutations in transgene, Accept not all cells with transgenes

Question 15 · A level Paper 3 · June 2018 · Q05.3 · 3 marks

05.3	1. siRNA binds to / destroys mRNA for CENP-W; 2. Prevents translation of CENP-W; 3. (After / as) CENP-W reduces **so** does tubulin production;	3	1. Reject if siRNA binds to gene / DNA 1. and 2. Context is important, siRNA acts on mRNA for CENP-W, **not** tubulin. 1. and 2. Ref. to CENP-W required **once** for MP1 and MP2 2. Accept reduces translation of CENP-W

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07.3	1. Both are more effective than the control; 2. Differences in the means not (likely to be) due to chance **OR** Significant difference (in effectiveness between both types); 3. (As) SDs do not overlap; 4. HBsAg (reduced), not zero **OR** Replication (reduced), not zero; 5. Not (investigated in) humans **OR** (Investigated in) mice; 6. shRNA (more effective as) 7.5% (of control) compared with 50% for lhRNA; 7. No indication of sample size/number; 8. Long term effects not known **OR** Side effects not known; 9. No statistical test to determine significance; 10. (Investigated) in vitro **OR** Not (investigated) in vivo;	5 max

Mark points 4 to 10 = 4 max.

1. Accept both (results) are below the control.

2. Reject 'results are significant'.

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Question 17 · A level Paper 3 · June 2018 · Q06.4 · 2 marks

06.4	(Species **S** because) no mark 1. Species **S** change of 990,000 (per gram of soil); 2. Species **T** change of 9,900 (per gram of soil); **OR** (Species **T** because) no mark 3. Species **S** has 99% change; 4. Species **T** has 9900% change;	2	Award MP1 and 2 **OR** MP3 and 4 1. and 2. Accept standard forms 9.9 x 10^5 and 9.9 x 10^3 for either Accept for 1 mark for 100 times greater in correct context with no other calculations shown

Question 18 · A level Paper 3 · June 2018 · Q06.5 · 4 marks

06.5	1. They didn't count bacteria / cells / population(s); 2. Copies / number of mRNA related to amount of enzyme / amoA produced / translated; 3. Don't know how much mRNA / amoA produced by each cell; 4. Don't know if amoA (mRNA / enzyme) is linked to cell division / growth (of population);	4	Ignore ref. to other factors / other named factors affecting growth 3. Accept some bacteria produce more mRNA / amoA than others 4. Amount of amoA does not show cell division / growth 4. Reject references to mitosis / meiosis

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