

AQA A-LEVEL • 7402

BIOLOGY



EXAM QUESTION PACK

3.8.4 RECOMBINANT DNA TECHNOLOGY

SCORE

/695

PERCENTAGE

%

STEMUP

These questions belong to AQA, not to us.

The examination questions, mark schemes and examiner reports in this pack were written and published by AQA. They are past assessment materials, reproduced here so that students can revise from them.

What we do not own

STEMUP is not affiliated with AQA and does not own, and makes no claim to own, any of the question, mark scheme or examiner report material in this pack. AQA has not endorsed, approved or been involved in the production of it. AQA and the qualification titles used here belong to their respective owners and appear for identification only.

Where to find the originals

AQA publishes its past papers, mark schemes and examiner reports free of charge. Always check the current specification at aqa.org.uk before you revise, because content and wording can change between series.

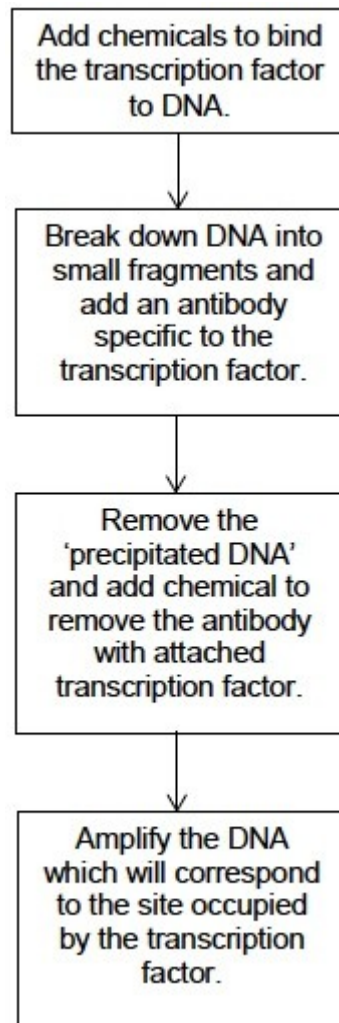
If you hold the rights to any material in this pack and would like it amended or removed, contact admin@stemup.co.uk and we will act on it.

Q1.

- (a) What is meant by a genome?

(1)

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



- (b) Explain why the antibody binds to the transcription factor.

(2)

- (c) Use the chart to explain what 'precipitated DNA' consists of.

(1)

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.

- (d) Suggest how single-stranded cDNA could prevent transcription of the *P34* gene.

(1)

- (e) Describe the roles of **two** named types of enzymes used to insert DNA fragments into plasmids.

Type of enzyme _____

Role _____

Type of enzyme _____

Role _____

(2)

- (f) The soybean cells were screened for the presence of the P34 protein. This process involved the use of gel electrophoresis to separate proteins extracted from soybean cells.

Suggest **two** features of the structure of different proteins that enable them to be separated by gel electrophoresis.

1. _____

2. _____

(2)
(Total 9 marks)

Q2.

Read the following passage.

Complete achromatopsia is a form of complete colour blindness. It is caused by having only rods and no functional cone cells. People with complete achromatopsia have difficulty in seeing detail. Complete achromatopsia is caused by an autosomal recessive allele and is usually very rare in populations with only one in 40 000 being affected. However on the Pacific island of Pingelap ten percent of the population are affected. 5

One form of red-green colour blindness is caused by a sex-linked recessive allele which affects more men than women. People with this red-green colour blindness are unable to distinguish between red and green, and also between other colours. They have green-sensitive cones but the photoreceptive pigment they contain does not function. 10

Scientists investigated the use of gene therapy to correct red-green colour blindness in monkeys. They injected viruses containing the gene for the green-sensitive pigment directly into the eyes of the monkeys. Although the monkeys maintained two years of colour vision, there is debate on whether this form of gene therapy is worthwhile. No clinical trials of this procedure have been carried out on humans. Current research into the treatment of red-green colour blindness involves the use of induced pluripotent stem cells (iPS cells). The use of iPS cells could have advantages over the use of gene therapy. 15
20

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

- (a) People with complete achromatopsia have difficulty in seeing detail (lines 2–3).

Explain why.

(3)

- (b) Ten percent of the population on the Pacific island of Pingelap are affected by complete achromatopsia (lines 3–6).

Use the Hardy-Weinberg equation to calculate the percentage of this population who are heterozygous for this disorder.
Show your working.

Answer = _____ %

(2)

- (c) Red-green colour blindness affects more men than women (lines 7–8).

Explain why.

(2)

- (d) People with red-green colour blindness are unable to distinguish between red and green, and also between other colours (lines 8–10).

Explain why.

(3)

- (e) Current research into the treatment of red-green colour blindness involves the use of induced pluripotent stem cells (iPS cells) (lines 17–19).

Suggest how iPS cells could correct red-green colour blindness.

(2)

- (f) The use of iPS cells could have advantages over the use of gene therapy to correct red-green colour blindness (lines 19–20).

Using the information from the passage, suggest and explain reasons why.

(3)

Q3.

Dengue is a serious disease that is caused by a virus. The virus is carried from one person to another by a mosquito, *Aedes aegypti*. One method used to try to reduce transmission of this disease is the Sterile Insect Technique (SIT). This involves releasing large numbers of sterile (infertile) male *A. aegypti* into the habitat. These males have been made infertile by using radiation.

- (a) Explain how using the SIT could reduce transmission of dengue.

(2)

- (b) Describe how the mark-release-recapture method could be used to determine the population of *A. aegypti* at the start of the investigation.

(3)

- (c) The release of radiation-sterilised *A. aegypti* has not been very successful in controlling the transmission of dengue.

Suggest **one** reason why.

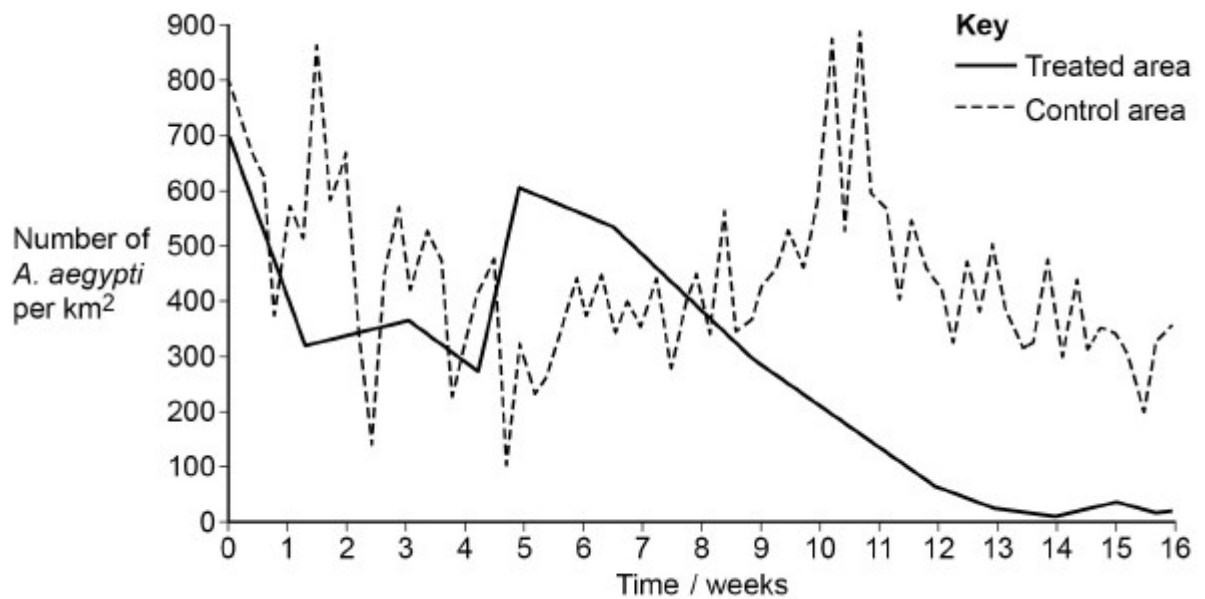
(1)

- (d) Recently a new method was developed to control *A. aegypti*. Scientists produced transgenic males carrying a 'lethal gene' which kills their offspring before they can reproduce.

The scientists released transgenic males every week in one area of a city in Brazil. At regular intervals they determined the number of *A. aegypti* per km² in the area where transgenic males were released and in a control area where no transgenic

males were released.

The graph shows their results.



Suggest why the scientists released more transgenic males every week.

(1)

- (e) The release of transgenic males proved successful in reducing the number of *A. aegypti*.

Describe how the results in the diagram above support this conclusion.

(2)

(Total 9 marks)

Q4.

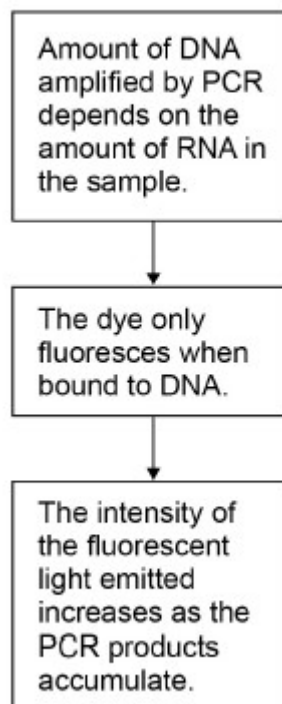
One way to detect and measure accurately the amount of RNA in a tissue sample is by RT-PCR (reverse transcriptase-polymerase chain reaction).

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 1**.

Figure 1



(a) Explain the role of reverse transcriptase in RT-PCR.

(1)

(b) Explain the role of DNA polymerase in RT-PCR.

(1)

(c) Any DNA in the sample is hydrolysed by enzymes before the sample is added to the

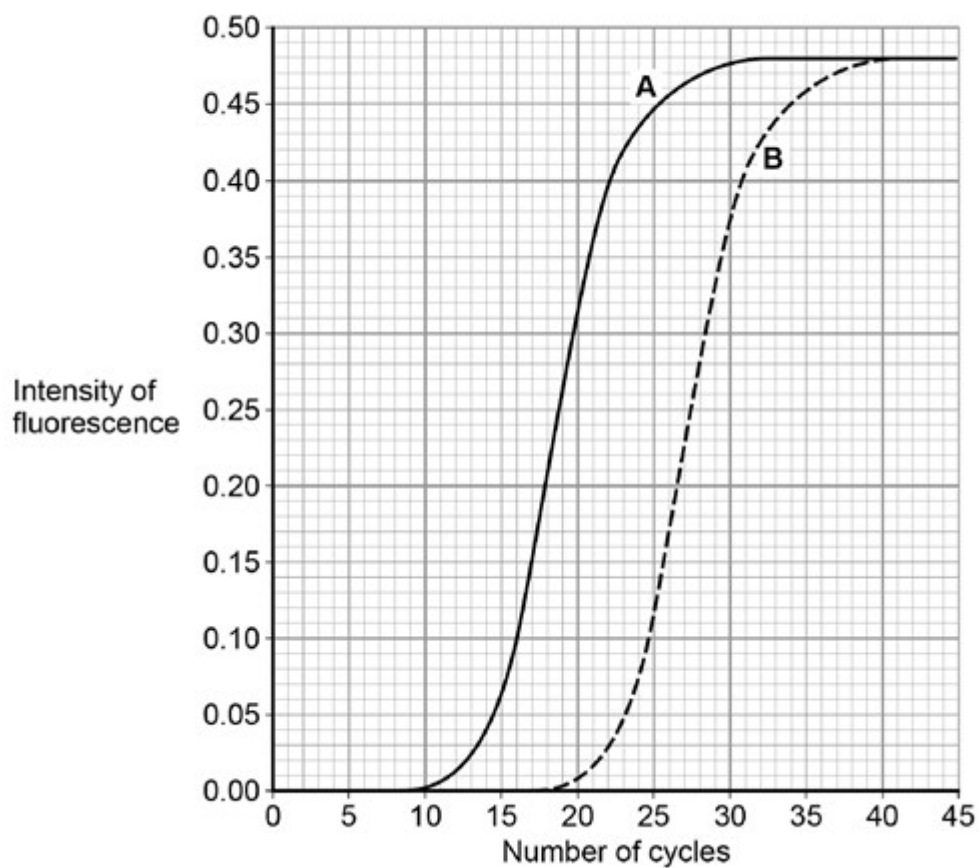
reaction mixture.

Explain why.

(2)

- (d) **Figure 2** shows the results from using RT-PCR to detect RNA in two different samples, **A** and **B**.

Figure 2



A quantitative comparison can be made of the amount of RNA in samples **A** and **B**. This involves determining the number of cycles required to reach 50% maximum concentration of DNA (**C**).

The amount of RNA in a sample can be measured as: $\frac{1}{C}$

Use this information to calculate the ratio for RNA content in sample **A** : RNA content in sample **B**.

(2)

- (e) Suggest **one** reason why DNA replication stops in the polymerase chain reaction.

(1)

- (f) 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)

(Total 9 marks)

Q5.

People suffering from pituitary dwarfism do not make enough human growth hormone (HGH). They can be treated using injections of HGH.

A geneticist wants to transform the bacterium, *Escherichia coli*, to make HGH by adding the gene coding for HGH.

The geneticist could obtain the *HGH* gene using any one of three methods.

1. Use restriction enzymes to cut out a fragment of DNA containing the *HGH* gene from a human genome.
2. Convert mRNA for HGH into cDNA using reverse transcriptase.
3. Create the *HGH* gene using a 'gene machine'.

- (a) The geneticist decided **not** to use restriction enzymes to cut out a fragment of DNA containing the *HGH* gene from a human genome. She made this decision because only methods 2 and 3 would produce DNA that *E. coli* could use to make HGH.

Explain why only methods 2 and 3 would produce DNA that *E. coli* could use to make HGH.

(2)

- (b) The geneticist concluded it would be faster to create the *HGH* gene using a gene machine than by using reverse transcriptase to convert mRNA for HGH into cDNA.

Suggest why the geneticist reached this conclusion.

(1)

- (c) After obtaining copies of the *HGH* gene, the geneticist will attempt to insert them into plasmid vectors.

Describe how the geneticist would attempt to insert copies of the *HGH* gene into these plasmids.

(3)

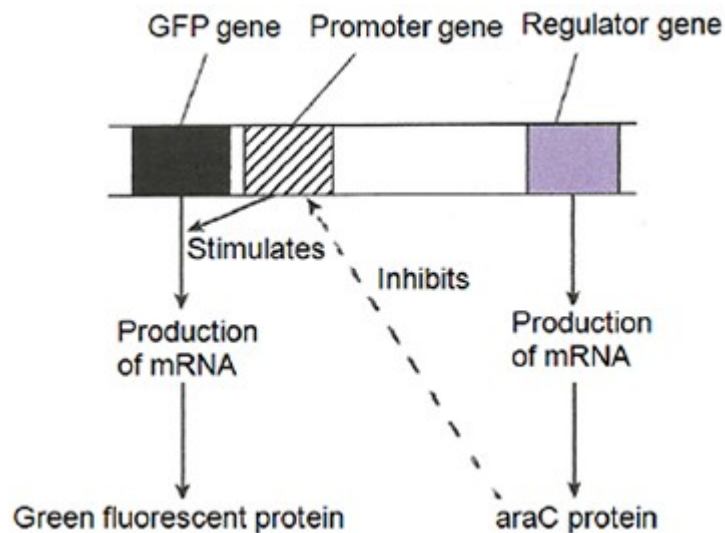
- (d) The geneticist plans to use the plasmids containing the *HGH* gene to try to transform cells of *E. coli*. She knows that some *E. coli* might not take up the plasmid.

To enable her to identify which bacteria have taken up the plasmid with the *HGH* gene, the plasmids she intends to use contain a gene that codes for a green fluorescent protein (GFP). Bacteria that contain this plasmid glow green under UV light.

Suggest **one** advantage of using this gene for GFP to identify bacteria that have taken up plasmids.

(1)

The diagram below shows part of the plasmid containing the gene that codes for GFP. It also shows the roles of two genes that control the GFP gene.



- (e) Arabinose is a sugar that can bind to the araC protein.

Use information in the diagram to suggest why the geneticist must include arabinose in the agar on which she hopes to grow *E. coli* containing the transgenic plasmids.

(2)

(Total 9 marks)

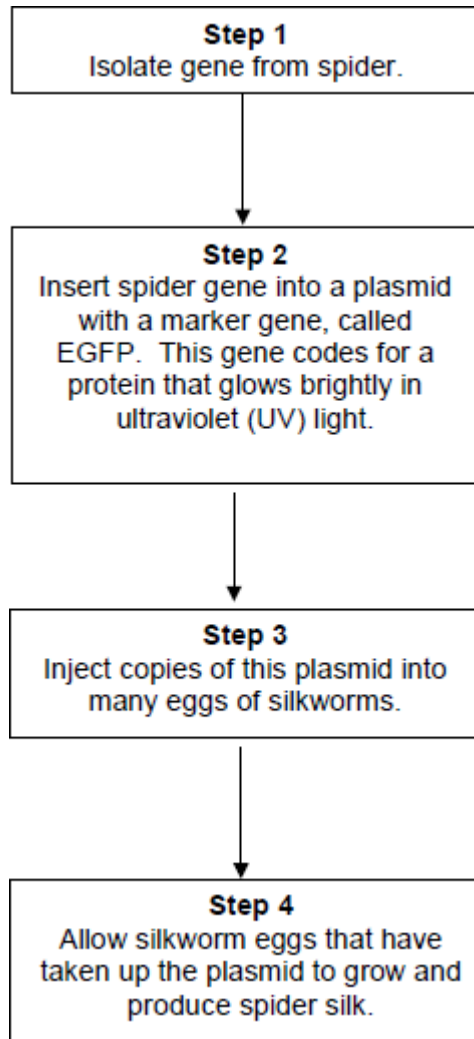
Q6.

Silkworms secrete silk fibres, which are harvested and used to manufacture silk fabric.

Scientists have produced genetically modified (GM) silkworms that contain a gene from a spider.

The GM silkworms secrete fibres made of spider web protein (spider silk), which is stronger than normal silk fibre protein.

The method the scientists used is shown in the figure below.



- (a) Suggest why the plasmids were injected into the eggs of silkworms, rather than into the silkworms.

(2)

- (b) Suggest why the scientists used a marker gene and why they used the EGFP gene.

(2)

The scientists ensured the spider gene was expressed only in cells within the silk glands.

- (c) What would the scientists have inserted into the plasmid along with the spider gene to ensure that the spider gene was only expressed in the silk glands of the silkworms?

(1)

- (d) Suggest **two** reasons why it was important that the spider gene was expressed only in the silk glands of the silkworms.

1.

2.

(2)

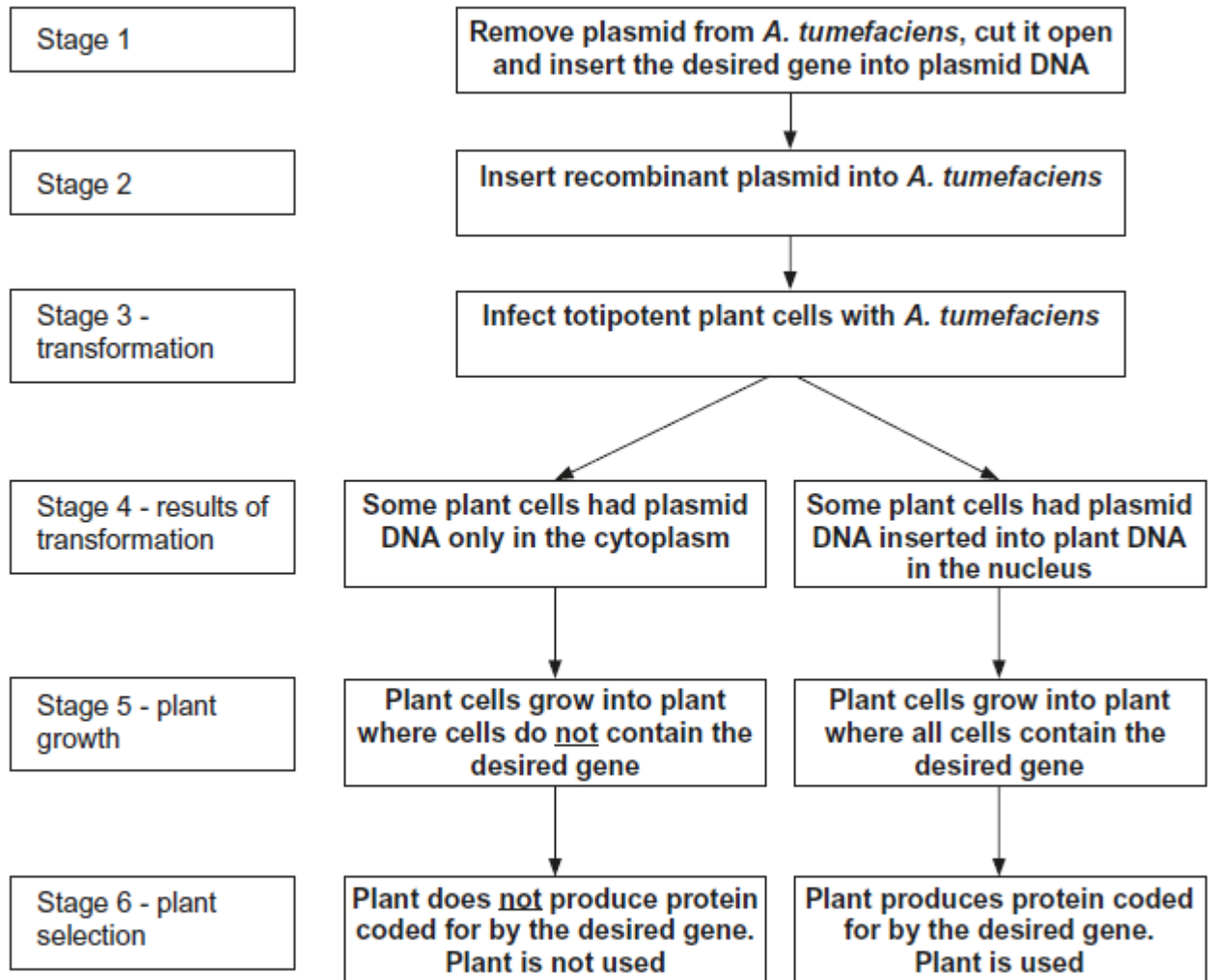
(Total 7 marks)

Q7.

Agrobacterium tumefaciens is a bacterium that is often used in recombinant DNA technology to produce transformed plants that benefit humans.

A. tumefaciens contains a plasmid which can be used as a vector to transfer a desired gene into plant cells. These plant cells may then develop into plants which produce the protein coded for by the desired gene.

The diagram outlines this process.



- (a) (i) In stage 1, an enzyme is used to cut open the plasmid.
Name the type of enzyme used to cut open the plasmid.

_____ (1)

- (ii) In stage 1, another enzyme is used to insert the desired gene into the plasmid DNA.
Name the type of enzyme used to insert the gene into the plasmid.

_____ (1)

- (b) In stage 4, some plant cells had plasmid DNA only in their cytoplasm. In other plant cells, the plasmid DNA had become inserted into plant DNA in the nucleus.

In stage 5, only cells with plasmid DNA inserted into the plant DNA in the nucleus grew into plants where all the cells contained the desired gene.

Explain why some of the plants in stage 5 contained the desired gene in all of their cells and others did not.

(3)

- (c) The **desired gene** in the diagram was from an insect. In stage 6, the plant containing this gene was able to use it to synthesise an insect protein.

The plant is able to synthesise the insect protein. Explain why this is possible.

(3)

(Total 8 marks)

Q8.

Some populations of flies are becoming resistant to insecticides intended to kill them.

Scientists developed a method for finding out whether a fly was carrying a recessive allele, **r**, that gives resistance to an insecticide. The dominant allele, **R**, of this gene does not give resistance.

The scientists:

- crossed flies with genotype **RR** with flies with genotype **rr**
- obtained DNA samples from the parents and offspring
- used the same restriction endonuclease enzymes on each sample, to obtain DNA fragments.

- (a) Explain why the scientists used the same restriction endonuclease enzymes on each DNA sample.

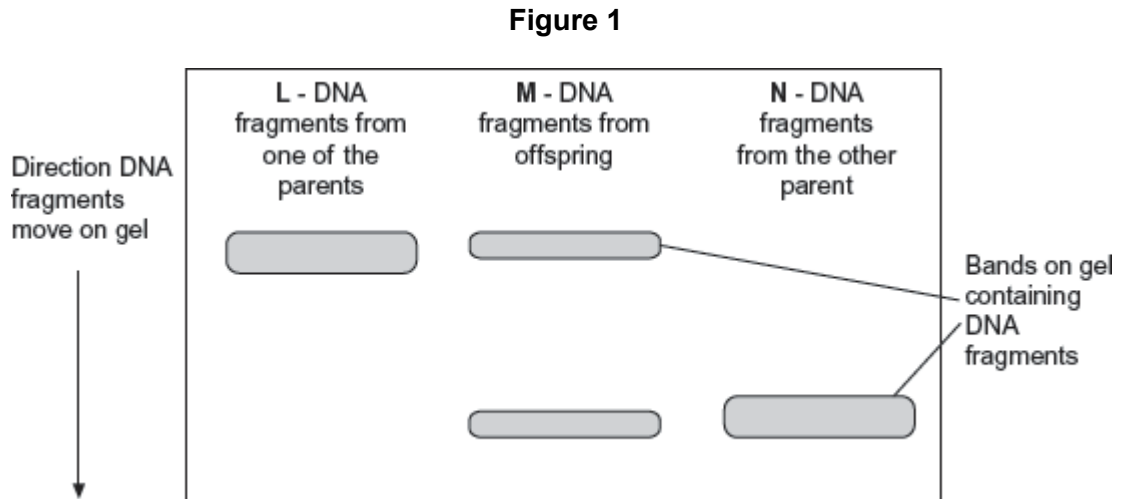
(2)

The scientists added two different primers to each sample of DNA fragments for the polymerase chain reaction (PCR).

- Primer A3 only binds to a 195 base-pair fragment from allele **r**.
- Primer A4 only binds to a 135 base-pair fragment from allele **R**.

The scientists separated the DNA fragments produced by the PCR on a gel where shorter fragments move further in a given time.

Their results are shown in **Figure 1**.



- (b) Explain why primer A3 and primer A4 only bind to specific DNA fragments.

(2)

- (c) Use all the information given to explain the results in **Figure 1**.

(3)

- (d) The scientists wanted to know on which chromosome the gene with alleles **R** and **r** was located. From the flies with genotype **RR**, they obtained cells that were in mitosis and added a labelled DNA probe specific for allele **R**. They then looked at the cells under an optical microscope.

Explain why they used cells that were in mitosis.

(2)

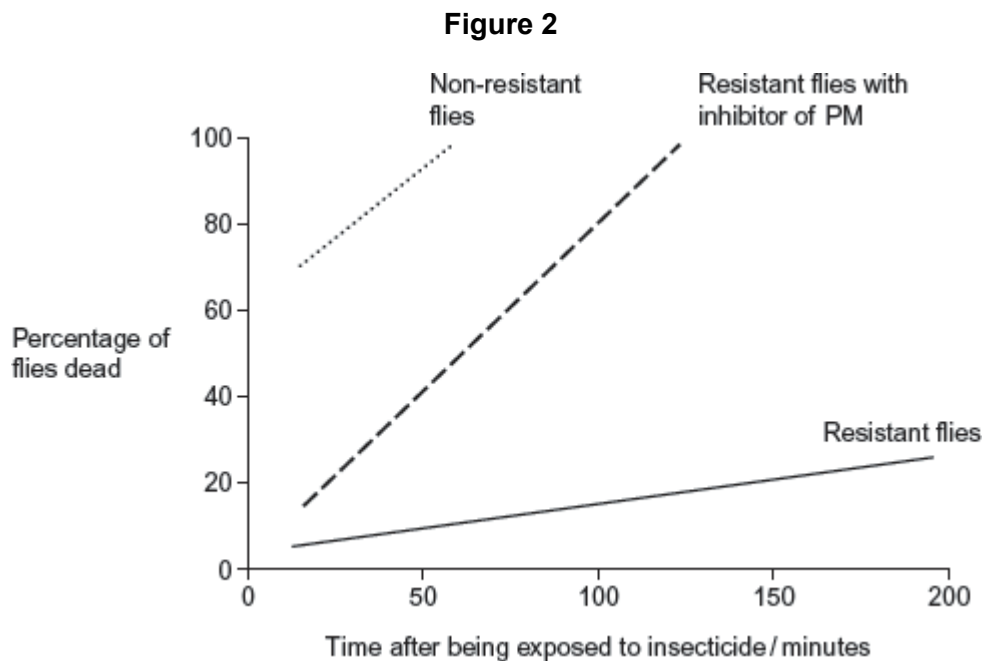
- (e) Another group of scientists thought that pesticide resistance in some flies was related to increased activity of an enzyme called P450 monooxygenase (PM). This enzyme breaks down insecticides.

The scientists obtained large numbers of resistant and non-resistant flies. They then set up the following experiments.

- Non-resistant flies exposed to insecticide.
- Resistant flies exposed to insecticide.
- Resistant flies treated with an inhibitor of PM and then exposed to insecticide.

They then determined the percentage of flies that were dead at different times after being exposed to insecticide.

Figure 2 shows their results.



- (i) Explain why the scientists carried out the control experiment with the non-resistant flies.

(2)

- (ii) The scientists concluded that the resistance of the flies to the insecticide is partly due to increased activity of PM but other factors are also involved.

Explain how these data support this conclusion.

(4)

(Total 15 marks)

Q9.

Scientists wanted to measure how much mRNA was transcribed from allele **A** of a gene in a sample of cells. This gene exists in two forms, **A** and **a**.

The scientists isolated mRNA from the cells. They added an enzyme to mRNA to produce cDNA.

- (a) Name the type of enzyme used to produce the cDNA.

(1)

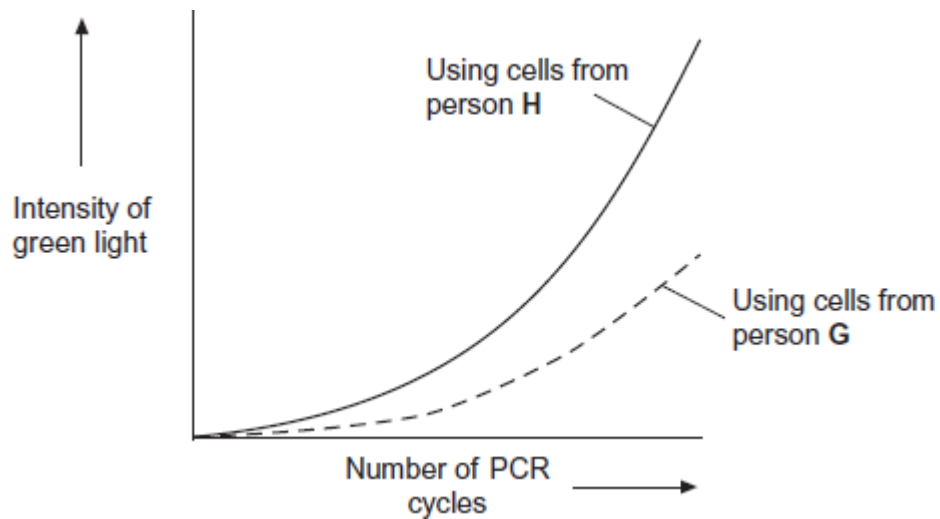
The scientists used the polymerase chain reaction (PCR) to produce copies of the cDNA. They added a DNA probe for allele **A** to the cDNA copies. This DNA probe had a dye attached to it. This dye glows with a green light **only** when the DNA probe is attached to its target cDNA.

- (b) Explain why this DNA probe will only detect allele **A**.

(2)

- (c) The scientists used this method with cells from two people, **H** and **G**. One person was homozygous, **AA**, and the other was heterozygous, **Aa**. The scientists used the PCR and the DNA probe specific for allele **A** on the cDNA from both people.

The figure shows the scientists' results.



- (i) Explain the curve for person H.

(3)

- (ii) Which person, H or G, was heterozygous, Aa? Explain your answer.

(2)

(Total 8 marks)

Q10.

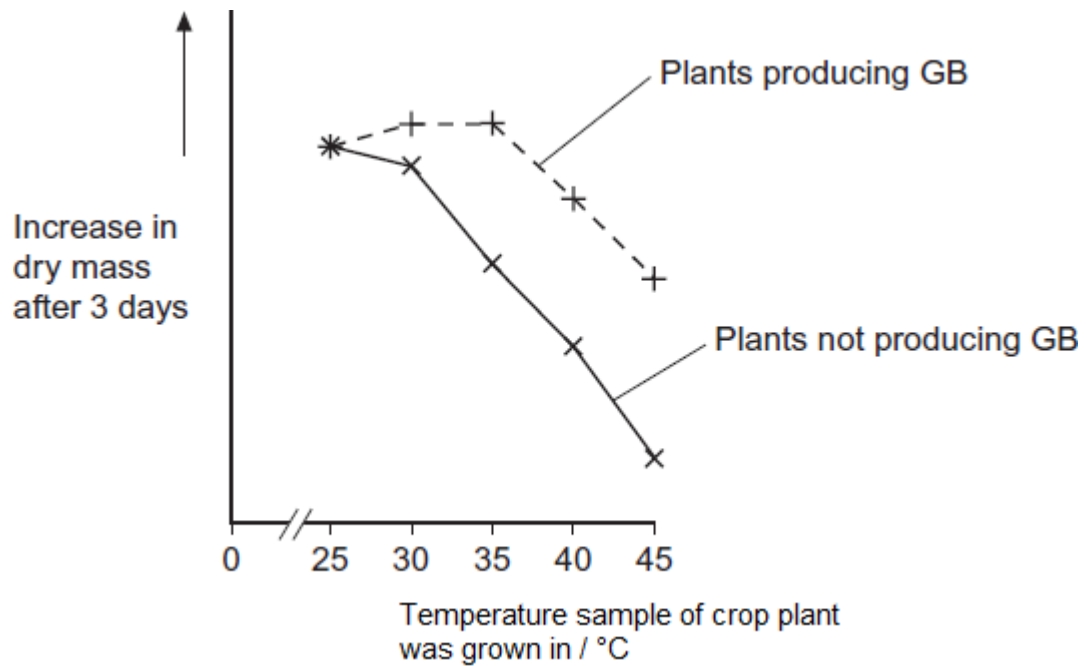
Some species of crop plant produce a substance called glycinebetaine (GB).

Scientists transferred the gene for GB into a species of crop plant that does not normally produce GB. These genetically modified plants then produced GB.

The scientists grew large numbers of the same crop plant with and without the gene at different temperatures. After 3 days, they found the increase in dry mass of the plants.

Figure 1 shows their results.

Figure 1



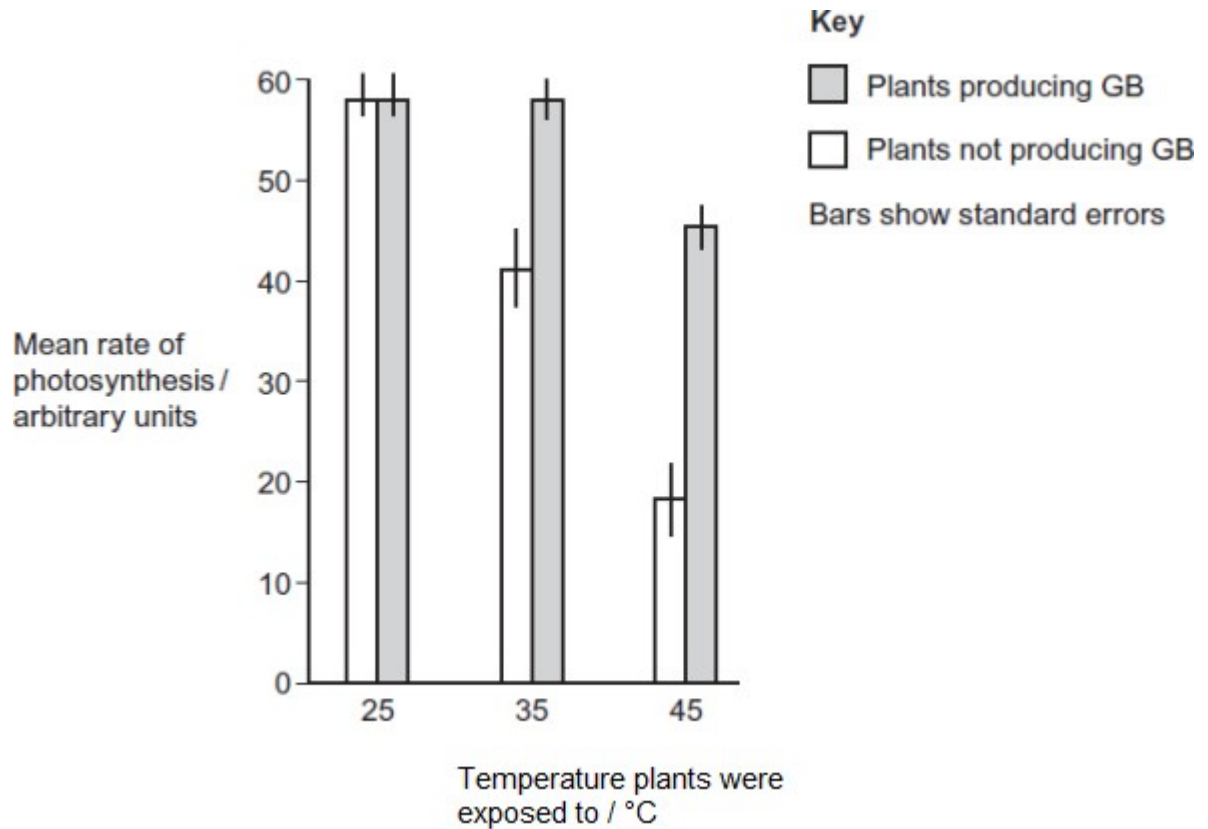
- (a) Describe the effect on growth of transferring the gene for GB into this plant.

(2)

- (b) The scientists measured the rate of photosynthesis in plants that produce GB and plants that do not produce GB at 25°C, 35°C and 45°C.

Figure 2 shows their results.

Figure 2



- (i) The scientists concluded that the production of GB protects photosynthesis from damage by high temperatures.

Use these data to support this conclusion.

(1)

- (ii) Use the data from **Figure 2** for plants that do not produce GB to explain the effect of temperature on changes in dry mass of the plants shown in **Figure 1**.

(4)

Rubisco activase is an enzyme found in chloroplasts. It activates the light-independent reaction of photosynthesis.

The scientists discovered that, as temperature increased from 25°C to 45°C, rubisco activase began attaching to thylakoid membranes in chloroplasts and this stopped it working.

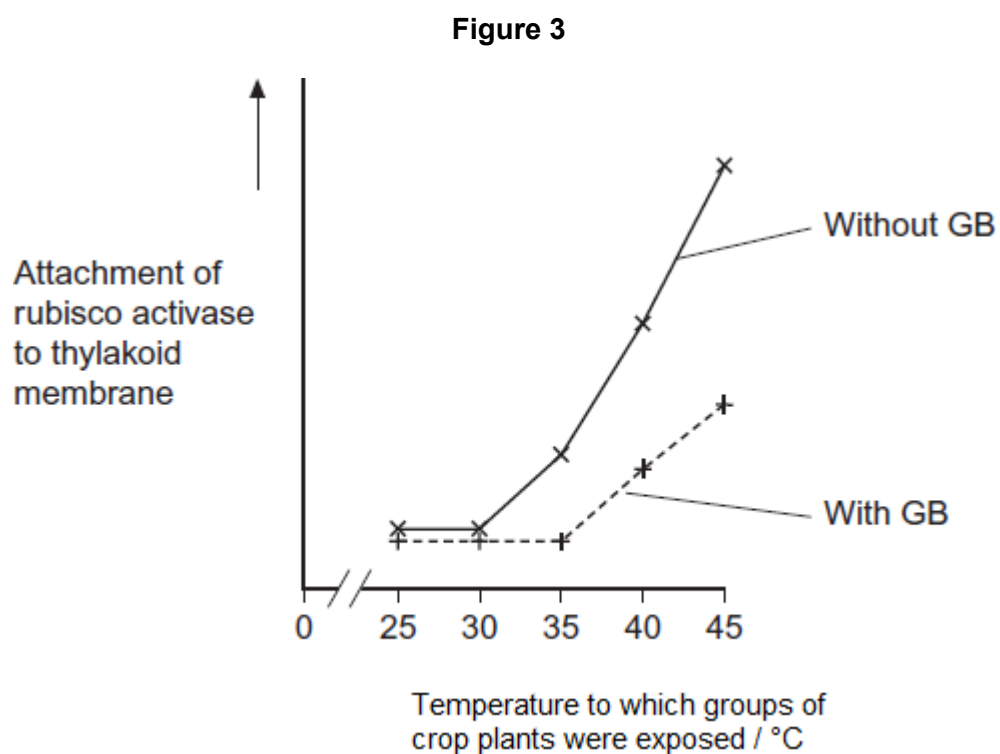
- (c) Rubisco activase stops working when it attaches to a thylakoid.

Use your knowledge of protein structure to explain why.

(2)

- (d) The scientists investigated the effect of GB on attachment of rubisco activase to thylakoid membranes at different temperatures.

Figure 3 shows their results.



Use information from **Figure 2** and **Figure 3** to suggest how GB protects the crop plant from high temperatures.

(4)

- (e) The scientists' hypothesis at the start of the investigation was that crop plants genetically engineered to produce GB would become more resistant to high environmental temperatures.
The scientists developed this hypothesis on the basis of previous research on crops that are grown in hot climates.

Suggest how the scientists arrived at their hypothesis.

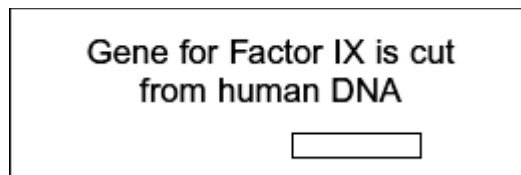
(2)

(Total 15 marks)

Q11.

Haemophilia is a genetic condition in which blood fails to clot. Factor IX is a protein used to treat haemophilia. Sheep can be genetically engineered to produce Factor IX in the milk produced by their mammary glands. The diagram shows the stages involved in this process.

Stage 1



Stage 2

Jellyfish gene is attached to
the human gene



Stage 3

Promoter DNA from
sheep is attached



Stage 4

Copies of this DNA are
inserted into the nuclei of
body cells from sheep



Stage 5

Each nucleus is
transplanted into a sheep
egg cell from which the
original nucleus has
been removed



Stage 6

The egg cells divide to form an embryo. Each embryo is implanted into the uterus of a different sheep

- (a) Name the type of enzyme that is used to cut the gene for Factor IX from human DNA (Stage 1).

_____ (1)

- (b) (i) The jellyfish gene attached to the human Factor IX gene (Stage 2) codes for a protein that glows green under fluorescent light. Explain the purpose of attaching this gene.

(2)

- (ii) The promoter DNA from sheep (Stage 3) causes transcription of genes coding for proteins found in sheep milk.

Suggest the advantage of using this promoter DNA.

(2)

- (c) Many attempts to produce transgenic animals have failed. Very few live births result from the many embryos that are implanted.

- (i) Suggest **one** reason why very few live births result from the many embryos that are implanted.

(2)

- (ii) It is important that scientists still report the results from failed attempts to produce transgenic animals. Explain why.

(2)

(Total 9 marks)

Q12.

- (a) Adrenaline binds to receptors in the plasma membranes of liver cells. Explain how this causes the blood glucose concentration to increase.

(2)

- (b) Scientists made an artificial gene which codes for insulin. They put the gene into a virus which was then injected into rats with type I diabetes. The virus was harmless to the rats but carried the gene into the cells of the rats.

The treated rats produced insulin for up to 8 months and showed no side-effects. The scientists measured the blood glucose concentrations of the rats at regular intervals. While the rats were producing the insulin, their blood glucose concentrations were normal.

- (i) The rats were not fed for at least 6 hours before their blood glucose concentration was measured. Explain why.

(1)

- (ii) The rats used in the investigation had type I diabetes. This form of gene therapy may be less effective in treating rats that have type II diabetes. Explain why.

(1)

- (iii) Research workers have suggested that treating diabetes in humans by this method of gene therapy would be better than injecting insulin. Evaluate this suggestion.

(4)

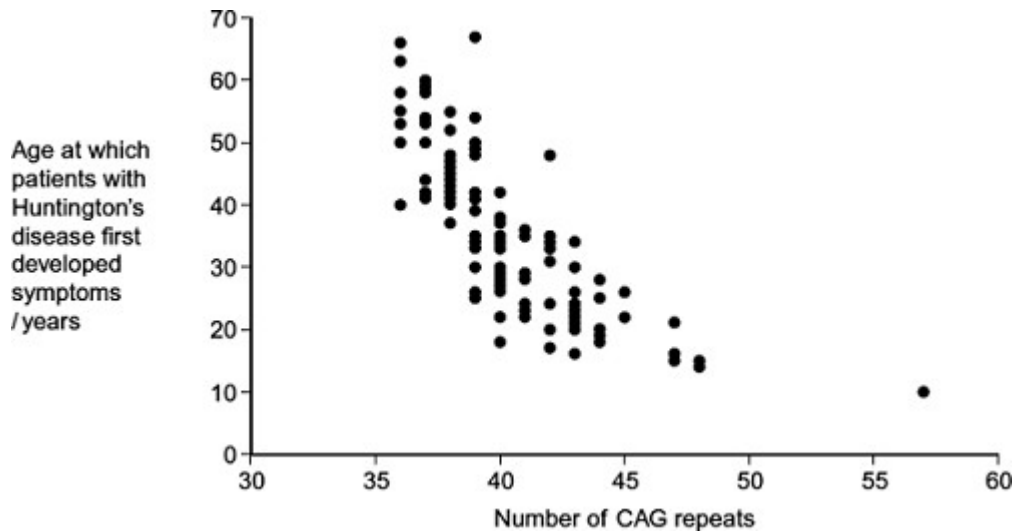
(Total 8 marks)

Q13.

Huntington's disease is a genetic condition that leads to a loss in brain function. The gene involved contains a section of DNA with many repeats of the base sequence CAG. The number of these repeats determines whether or not an allele of this gene will cause Huntington's disease.

- An allele with 40 or more CAG repeats will cause Huntington's disease.
- An allele with 36 – 39 CAG repeats may cause Huntington's disease.
- An allele with fewer than 36 CAG repeats will not cause Huntington's disease.

The graph shows the age at which a sample of patients with Huntington's disease first developed symptoms and the number of CAG repeats in the allele causing Huntington's disease in each patient.



- (a) (i) People can be tested to see whether they have an allele for this gene with more than 36 CAG repeats. Some doctors suggest that the results can be used to predict the age at which someone will develop Huntington's disease.

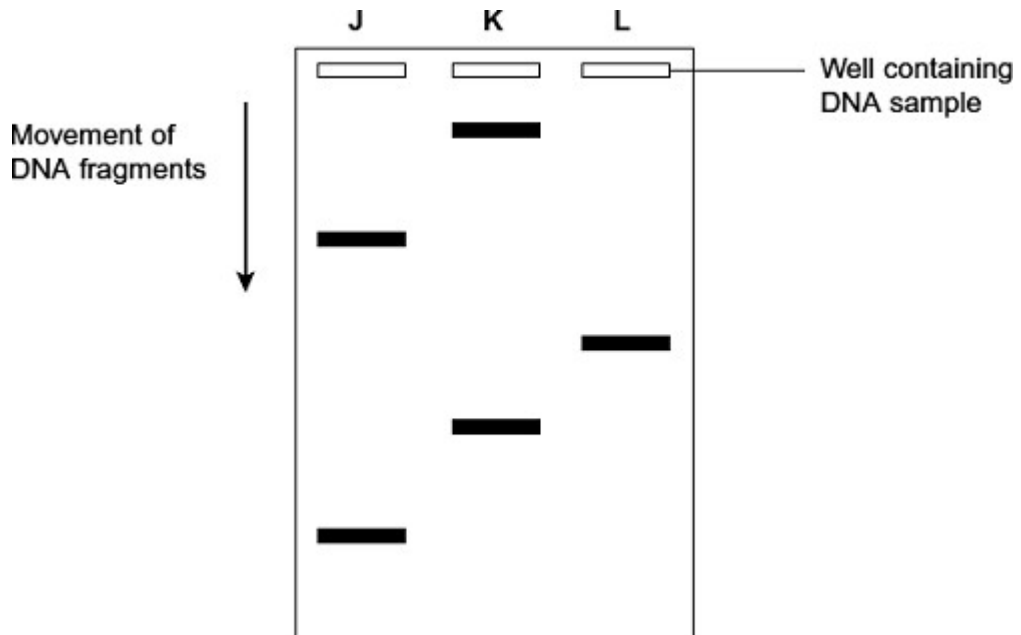
Use information in the graph to evaluate this suggestion.

(3)

- (ii) Huntington's disease is always fatal. Despite this, the allele is passed on in human populations. Use information in the graph to suggest why.

(2)

- (b) Scientists took DNA samples from three people, **J**, **K** and **L**. They used the polymerase chain reaction (PCR) to produce many copies of the piece of DNA containing the CAG repeats obtained from each person. They separated the DNA fragments by gel electrophoresis. A radioactively labelled probe was then used to detect the fragments. The diagram shows the appearance of part of the gel after an X-ray was taken. The bands show the DNA fragments that contain the CAG repeats.



- (i) Only one of these people tested positive for Huntington's disease. Which person was this? Explain your answer.

Person _____

Explanation _____

(2)

- (ii) The diagram only shows part of the gel. Suggest how the scientists found the number of CAG repeats in the bands shown on the gel.

(1)

- (iii) Two bands are usually seen for each person tested. Suggest why only one band was seen for Person L.

(1)

(Total 9 marks)

Q14.

- (a) Scientists can use protein structure to investigate the evolutionary relationships between different species. Explain why.

(2)

- (b) Comparing the base sequence of genes provides more evolutionary information than comparing the structure of proteins. Explain why.

(2)

(Total 4 marks)

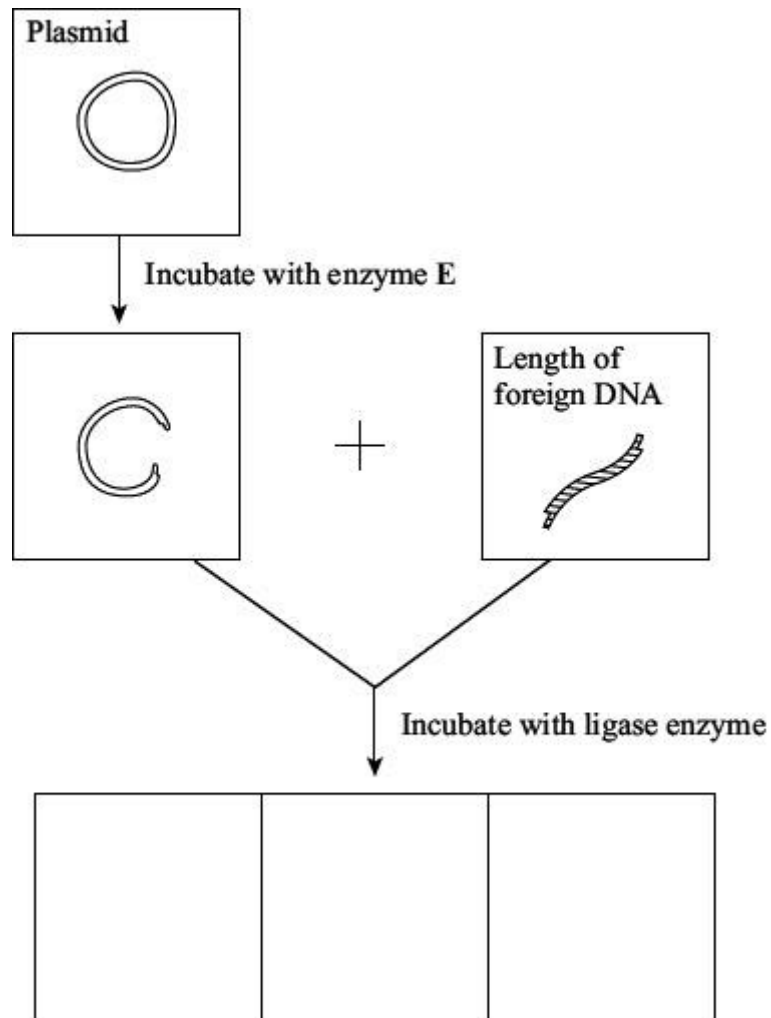
Q15.

Write an essay on using DNA in science and technology.

(Total 25 marks)

Q16.

Plasmids can be used as vectors to insert lengths of foreign DNA into bacteria. The diagram shows how this is achieved.



- (a) Name enzyme **E**.

(1)

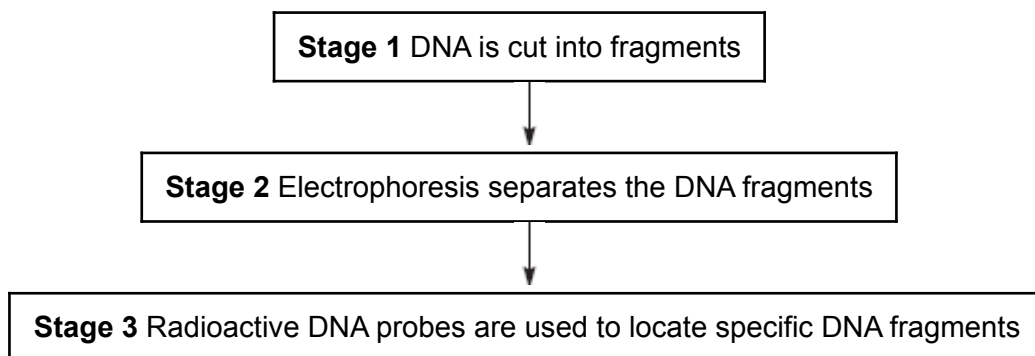
- (b) Cut plasmids and lengths of foreign DNA can join. What features of their ends allows them to join?

(2)

- (c) Draw **three** different structures that could be formed by incubating cut plasmids and lengths of foreign DNA with ligase. Use the spaces provided on the diagram.

Q17.

DNA probes may be used to identify the presence of specific genes associated with human diseases. The flow chart summarises the way in which they are used.



- (a) Name the enzyme used in **Stage 1**.

_____ (1)

- (b) Explain how electrophoresis separates the fragments of DNA in **Stage 2**.

_____ (2)

- (c) (i) What is a *DNA probe*?

_____ (2)

- (ii) Explain why *radioactive* DNA probes are used to locate specific DNA fragments.

Q18.

Read the following passage.

Herpes viruses cause cold sores and, in some cases, genital warts. Scientists are well on the way to producing an antibody which will counteract herpes infection. This antibody works by sticking to the virus and blocking its entry into cells. It has proved very effective in animal tests.

- 5 One drawback with this approach, however, is that antibodies are at present produced using hamster ovary cells. This method is expensive and only produces limited amounts. A new technique is being developed to produce antibodies from plants. It involves introducing the DNA which codes for the required antibody into crop plants such as maize.

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

- (a) (i) What is an antibody?

(2)

- (ii) Describe how antibodies are produced in the body following a viral infection.

(6)

- (b) Describe how the antibody gene could be isolated from an animal cell and introduced into a crop plant such as maize (lines 7-8).

(4)

- (c) Taking a course of these antibodies from plants to treat a herpes infection would not produce long-term protection against disease. Explain why.

(2)

- (d) Explain **one** advantage of using antibodies from plants to treat a disease, rather than antibodies produced in an experimental animal (lines 5-6).

(1)

(Total 15 marks)

Q19.

Scientists are working to produce a genetically modified bacterium to treat patients suffering from a disease of the digestive system. They plan to collect mRNA from human cells. This will be used to produce the DNA of the gene for the protein interleukin. They will then transfer this human gene into the bacterium *Lactococcus*. The scientists intend patients to swallow the genetically modified bacteria. These bacteria will release interleukin inside the digestive system to treat the disease.

- (a) (i) Name the type of enzyme which will be used to produce the DNA from the mRNA.

(1)

- (ii) It is easier to obtain the interleukin gene from mRNA rather than directly from the DNA removed from human cells. Explain why.

(1)

- (b) The scientists propose to put the gene directly into the DNA of *Lactococcus*. Describe the role of the enzyme ligase in this process.

(1)

(Total 3 marks)

Q20.

A gene was broken into fragments using enzyme **Z**. The mixture of fragments produced was then separated by electrophoresis.

- (a) What type of enzyme is enzyme **Z**?

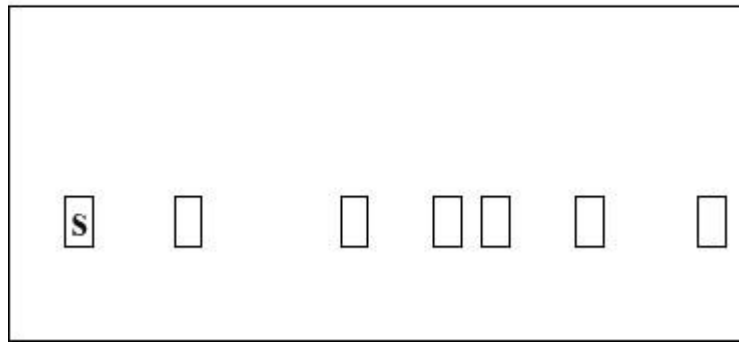
(1)

The table shows the number of base pairs present in the fragments.

Fragment	Number of base pairs ($\times 10^3$)
1	4.65
2	5.72
3	10.71
4	2.39
5	5.35
6	7.53

The diagram shows the electrophoresis gel used. The mixture of fragments was placed at the start point marked **S** and the process started. The boxes indicate the positions reached by the different fragments.

Direction of movement of fragments



- (b) Explain why base pairs are a suitable way of measuring the length of a piece of DNA.

(2)

- (c) (i) Write **6** above the appropriate box on the diagram to show the position you would expect fragment **6** to have reached.

(1)

- (ii) Explain how you arrived at your answer.

(1)

- (d) Enzyme **Z** recognises a particular sequence of bases in the gene. How many times does this sequence appear in the DNA of this gene?

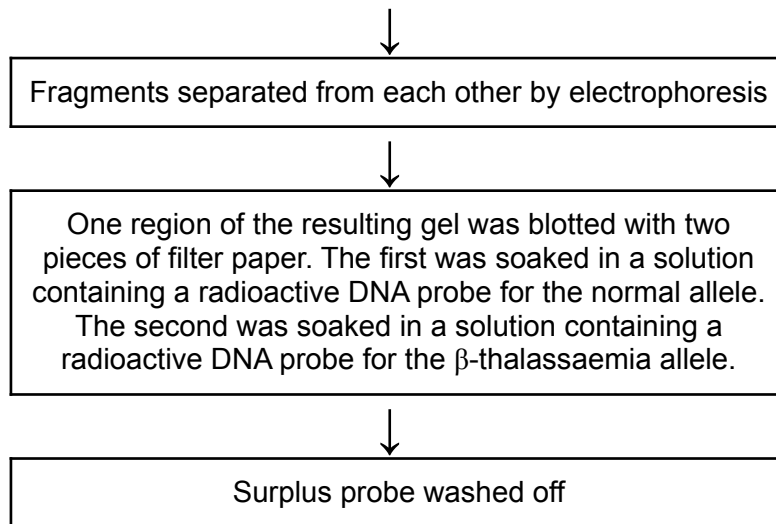
(1)

(Total 6 marks)

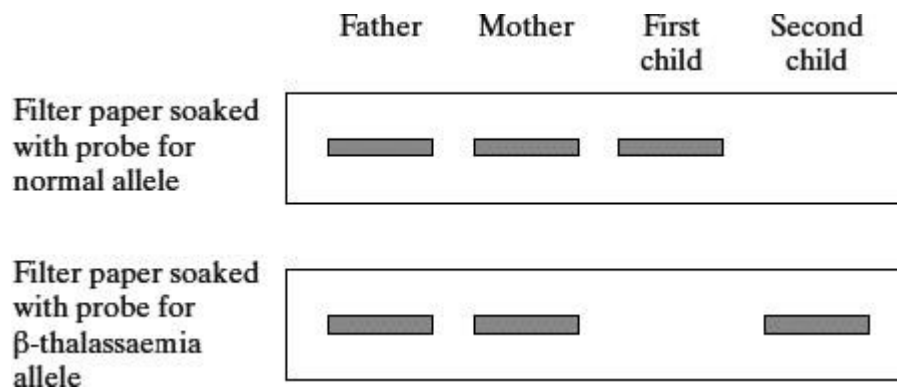
Q21.

β -thalassaemia is a genetic condition in which abnormal haemoglobin is produced. In one form, the recessive allele for β -thalassaemia, **t**, differs from the normal allele, **T**, by a single base-pair. A radioactive DNA probe was used to investigate the genotypes of four members of one family. The flowchart summarises the technique involved.

DNA samples extracted and cut into fragments using a restriction enzyme



The diagram below shows the appearance of the two pieces of filter paper which resulted from the investigation.



- (a) What is the probability that the next child that this couple have is a girl who has β -thalassaemia? Explain your answer.

(3)

- (b) (i) The fragment of DNA containing the normal allele and the fragment with the β -thalassaemia allele moved the same distance on the gel. Explain why.

(2)

- (ii) The allele for β -thalassaemia differs from the normal allele by only one base-pair. Explain why the probe used to identify these alleles consists of a piece of DNA twenty bases in length and not just one base.

(2)

(Total 7 marks)

Q22.

- (a) (i) Some human DNA was cut into separate pieces using a restriction enzyme which produced a staggered cut. A scientist wanted to insert these pieces of DNA into plasmids and used the same restriction enzyme to cut the plasmids. Explain why the pieces of human DNA would be able to join to the cut DNA of the plasmids.

(2)

- (ii) Which other enzyme must the scientist have added to the mixture to form recombinant plasmids?

(1)

- (b) A plasmid may be used as a vector. Explain what is meant by a *vector*.

(2)

- (c) Molecular biologists often use plasmids which contain antibiotic resistance genes. Explain the reason for this.

(2)
(Total 7 marks)

Q23.

Read the following passage.

Shark-fin soup is an expensive delicacy. To provide the basic ingredient, fishermen catch the sharks, hack the fins off and throw the dead bodies back into the ocean. But sharks are slow to mature and produce only a few offspring at a time, so they are vulnerable to overfishing. Monitoring the shark-fin trade is difficult, as once a fin has been cut off, it can be extremely difficult to work out precisely from which species it was taken.

The DNA from different species of sharks shows some differences in base sequence. This has enabled a new genetic fingerprinting technique to be developed. This technique would allow conservationists and fisheries managers to assess which of the 400 shark species are most threatened by the trade in shark fins.

10 An identification process has been developed using a range of “primers”. These are short pieces of single-stranded DNA that are complementary to a particular sequence of DNA. Each primer is specific to the DNA of one shark species.

15 The primers are added to DNA taken from a shark’s fin and the polymerase chain reaction is carried out. Only two primers, one at each end of a certain piece of DNA, will bind. The piece of DNA between the primers is replicated by the polymerase chain reaction. The primers that bind are specific to a particular species of shark and the length of the DNA fragment replicated differs for each species. When this DNA is run in an electrophoresis gel it produces a single band, enabling the researchers to identify which species of shark is involved.

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

- (a) (i) Explain why the DNA for each species of shark shows differences in base sequence (line 6).

(2)

- (ii) Each primer is specific to the DNA of one shark species (line 12).

Explain why a particular primer will only bind to the DNA of one species.

(2)

- (iii) The length of the replicated DNA fragment is different for each species.
Explain why this is important in identifying the shark species involved.

(3)

- (b) In conventional DNA fingerprinting, a series of bands is produced on the electrophoresis gel, resembling the rungs of a ladder. When the DNA in this new genetic fingerprinting technique is run in an electrophoresis gel it produces just one of these 'rungs'.

Explain the reason for the difference in the number of 'rungs' produced.

(2)

- (c) Describe the polymerase chain reaction.

(6)
(Total 15 marks)

Q24.

- (a) (i) Some human DNA was cut into separate pieces using a restriction enzyme which produced a staggered cut. A scientist wanted to insert these pieces of DNA into plasmids and used the same restriction enzyme to cut the plasmids. Explain why the pieces of human DNA would be able to join to the cut DNA of the plasmids.

(2)

- (ii) Which other enzyme must the scientist have added to the mixture to form recombinant plasmids?

(1)

- (b) A plasmid may be used as a vector. Explain what is meant by a *vector* in this context.

(2)

- (c) Molecular biologists often use plasmids which contain antibiotic resistance genes. Explain the reason for this.

(2)

5

Other cancer-detecting techniques involve removing a tissue sample from a patient. The tissue sample is used to obtain mRNA. By examining the mRNA, scientists can discover whether cancer is present.

(a) Describe how altered DNA may lead to cancer.

[illegible]

(b) Explain why fragments of DNA from cancer cells may be present in blood plasma (lines 3-4).

Page 44 of 102

- (c) Explain why the nucleic acid on the test strip will only bind to altered DNA (lines 4-5).

(2)

- (d) This test strip will allow cancers to be detected at a very early stage. Explain why cancer is more likely to be treated successfully if the disease is detected at a very early stage.

(2)

- (e) Explain how examining mRNA (line 7) enables scientists to discover whether cancer is present.

(3)

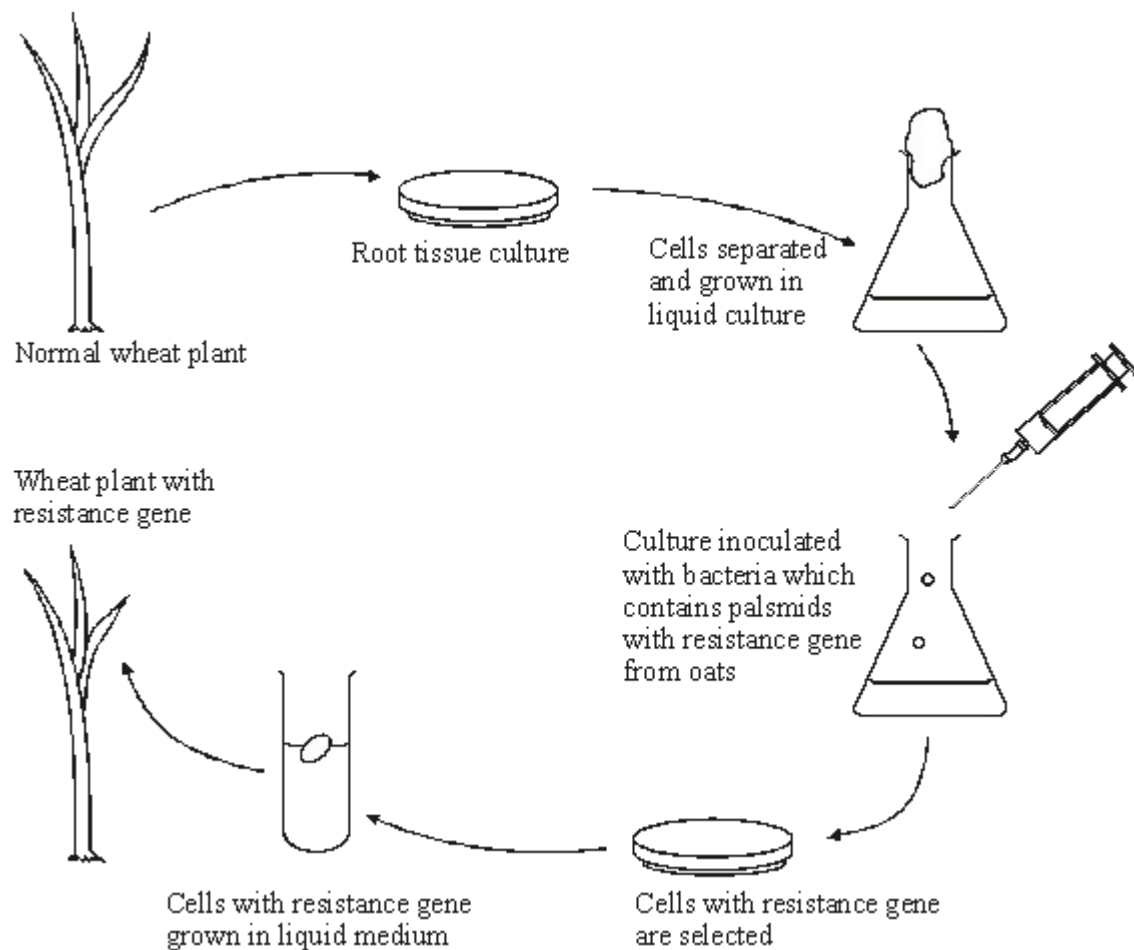
(Total 15 marks)

Q26.

'Take-all' is a disease of wheat caused by a fungus. It can cause serious damage to the crop.

There is no gene for resistance to this fungus in wheat. There is, however, a gene for resistance to this fungus present in oats.

The diagram shows how this gene might be transferred to wheat.



- (a) (i) The wheat plant with the resistance gene contains recombinant DNA. What is *recombinant DNA*?

(1)

- (ii) The plasmids act as vectors for the resistance gene. What is a *vector*?

(1)

- (iii) Suggest how cells with the resistance gene might be selected.

(2)

- (b) A laboratory has oat plants containing the resistance gene and a supply of plasmids.

This image shows a blank sheet of white paper with horizontal ruling lines. The lines are evenly spaced and run across the width of the page. There are no margins, text, or other markings on the paper.

(Total 10 marks)

Read the following passage.

5 The first half of the twentieth century was a time of hope for malarial control. The drugs
chloroquine and proguanil had just been discovered and there seemed a real possibility of a
malaria-free world. Unfortunately, this honeymoon ended almost as soon as it had started,
with the emergence of drug-resistant parasite populations. Scientists now accept that whatever
new drug they come up with, it is likely to have a very limited effective life. As a result, they
10 are increasingly looking at combinations of drugs.

Another, possibly more promising, approach has been the development of a DNA-based vaccine. In theory, all that is required is to identify the DNA from the parasite which encodes key antigens. Unfortunately, scientists have hit snags. Although they have succeeded in sequencing the human genome, the genome of the malarial parasite has created major difficulties. This is partly because of the very high proportion of the bases adenine and thymine. In some places these two bases average 80%, and on chromosomes 2 and 3 nearly 100% of the bases present are adenine and thymine. Because of this, it has proved impossible

- 25 to cut the relevant DNA with the commonly available restriction enzymes into pieces of a suitable size for analysis.

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

- (a) Explain how a resistant parasite population is likely to arise and limit the life of any new anti-malarial drug (lines 8 - 9).

(3)

- (b) A person has a 1 in 500 probability of being infected by a chloroquine-resistant strain of malarial parasite and a 1 in 500 probability of being infected by a proguanil-resistant strain. Use a calculation from these figures to explain why scientists are “increasingly looking at combinations of drugs” (lines 9 - 10).

(2)

- (c) (i) Explain why trials of the SPf66 vaccine needed a control.

(1)

- (ii) The controls for the SPf66 vaccine trials were considered inappropriate (line 17).

Suggest how the control groups in these trials should have been treated.

(2)

- (d) In some of the DNA of a malarial parasite, the proportion of adenine and thymine

bases averages 80% (lines 22 - 23). In this DNA what percentage of the nucleotides would you expect to contain

(i) phosphate; _____

(ii) guanine? _____

(2)

- (e) (i) Use your knowledge of enzymes to explain why restriction enzymes only cut DNA at specific restriction sites.

(3)

- (ii) Restriction enzymes that can cut the DNA of chromosomes 2 and 3 produce pieces that are too small for analysis. Explain why these restriction enzymes produce small DNA fragments.

(2)

(Total 15 marks)

Q28.

- (a) Plasmids are often used as vectors in genetic engineering.

- (i) What is the role of a vector?

(1)

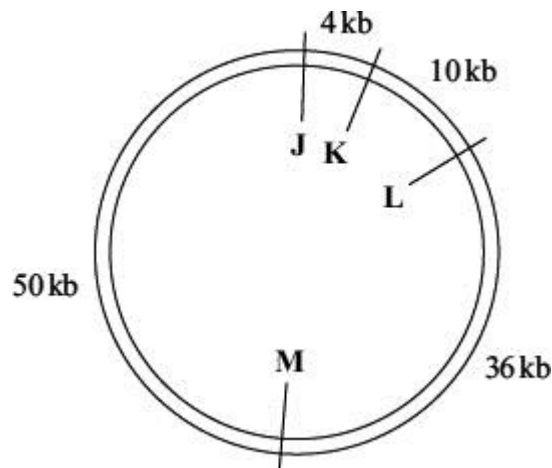
- (ii) Describe the role of restriction endonucleases in the formation of plasmids that contain donor DNA.

(2)

- (iii) Describe the role of DNA ligase in the production of plasmids containing donor DNA.

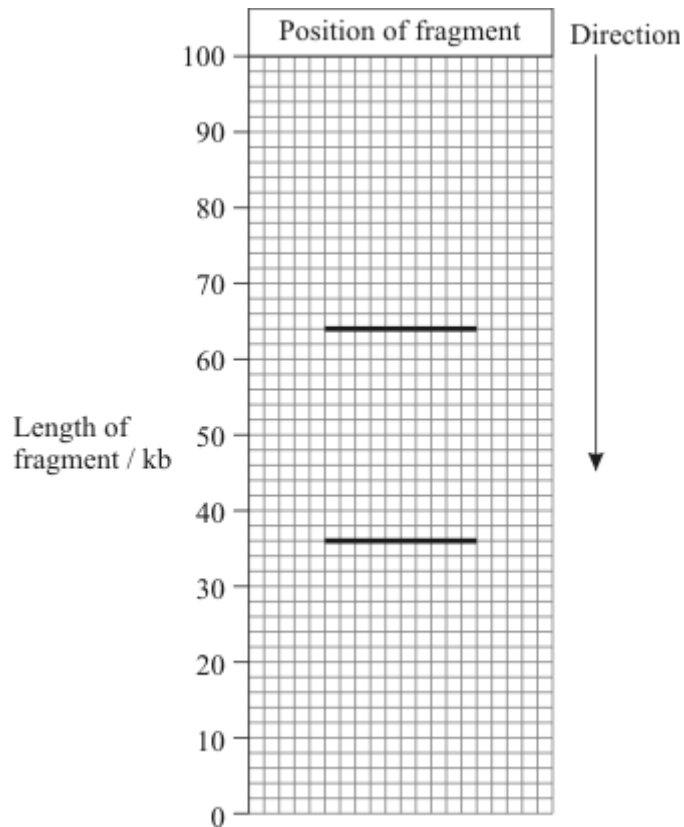
(1)

- (b) There are many different restriction endonucleases. Each type cuts the DNA of a plasmid at a specific base sequence called a restriction site. The diagram shows the position of four restriction sites, **J**, **K**, **L** and **M**, for four different enzymes on a single plasmid. The distances between these sites is measured in kilobases of DNA.



1 kb = 1 kilobase

The plasmid was cut using only two restriction endonucleases. The resulting fragments were separated by gel electrophoresis. The positions of the fragments are shown in the chart below.



- (i) Which of the restriction sites were cut?

(1)

- (ii) Explain your answer.

(1)

(Total 6 marks)

Q29.

- (a) An antigen in a vaccine leads to the production of antibodies. Describe the part played by B lymphocytes in this process.

(4)

- (b) Hepatitis B vaccine contains a viral antigen produced by genetically modified bacteria. Describe how the isolated gene that codes for a protein in the virus's coat could be transferred to the bacterial cells.

(3)

(Total 7 marks)

Q30.

A protein produced by a species of bacterium is toxic to caterpillars. The gene coding for this protein was removed and transferred into a crop plant.

- (a) (i) Describe how the gene could have been removed from the bacterial DNA.

(2)

- (ii) Many copies of the isolated gene were required. Name the process used in a laboratory to produce many copies of DNA from a small amount.

(1)

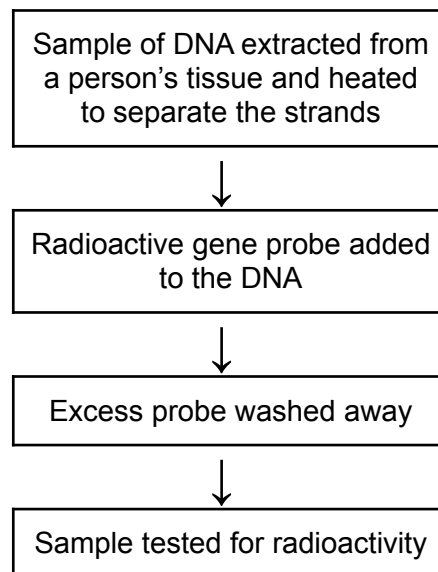
- (b) The gene was injected into isolated cells from the crop plant. These cells were then cloned and new plants grown from the cloned cells. Explain the advantage of inserting the gene into isolated plant cells rather than directly into cells within a whole plant.

(3)

(Total 6 marks)

Q31.

- (a) Cystic fibrosis can be caused by any one of several mutant alleles of the cystic fibrosis gene. The most common of these mutant alleles accounts for about 70% of cases of cystic fibrosis. The use of gene probes can identify individuals carrying this allele. Gene probes are single strands of DNA which are radioactively labelled. They have a base sequence that is complementary to a mutant allele. The main stages in using a gene probe are shown in the diagram.



Using the information given, explain how the use of a gene probe could enable the presence of a mutant allele of the cystic fibrosis gene to be detected.

(4)

- (b) Sheep have been genetically engineered to produce alpha-1-antitrypsin which is used to treat cystic fibrosis. Use your knowledge of this process to explain **one** argument for and **one** against using sheep in this way.

For

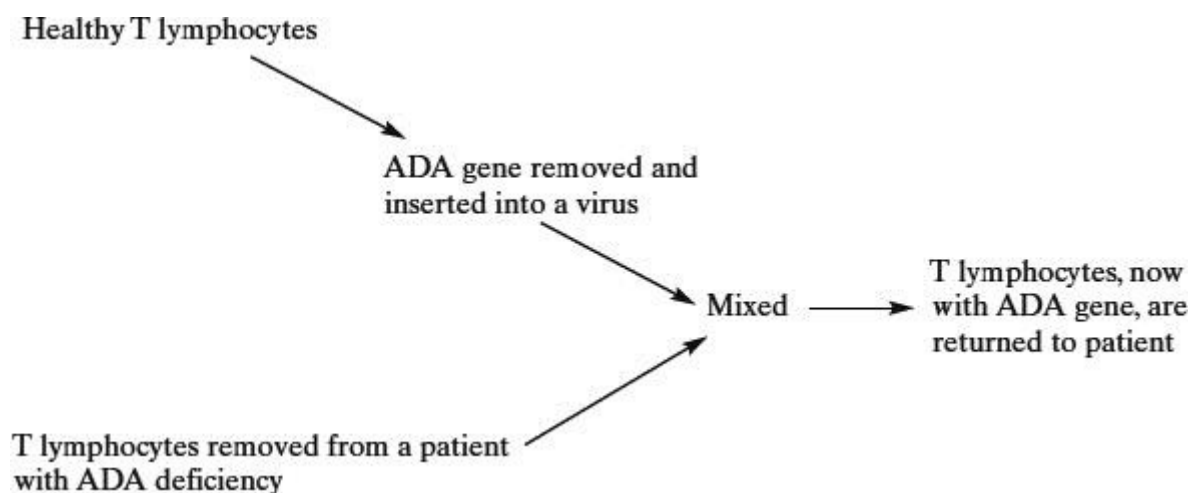
Against

(2)

(Total 6 marks)

Q32.

Gene therapy is used to treat the genetic disorder, ADA deficiency. Affected individuals are unable to produce the enzyme adenosine deaminase (ADA). Without this enzyme, T lymphocytes, a type of white blood cell, cannot provide immunity to infection. The diagram shows the processes involved in the treatment of ADA deficiency by gene therapy.



(a) What is meant by *gene therapy*?

(1)

(b) The ADA gene is inserted into a virus. Give **two** advantages of using a virus in gene therapy.

1. _____

2. _____

(2)

(c) Individuals who have been treated by this method of gene therapy do not pass on the ADA gene to their children. Explain why.

(1)

- (d) T lymphocytes are produced in bone marrow. A bone marrow transplant from a genetically matched donor can provide a permanent cure for ADA deficiency.

- (i) Suggest why bone marrow for a transplant is obtained from a genetically matched donor.

(1)

- (ii) Explain why treatment of ADA deficiency by gene therapy must be repeated at regular intervals, whereas a single bone marrow transplant can provide a permanent cure.

(2)

(Total 7 marks)

Q33.

Research scientists can increase the nutritional value of potatoes by genetically engineering potato plants. A gene which results in increased protein production has been removed from cells of an amaranth plant and inserted into cells of a potato plant.

- (a) Describe how a gene could be removed from cells of an amaranth plant and inserted into cells of a potato plant.

(6)

-
-
-
-
-
-

(2)

(Total 8 marks)

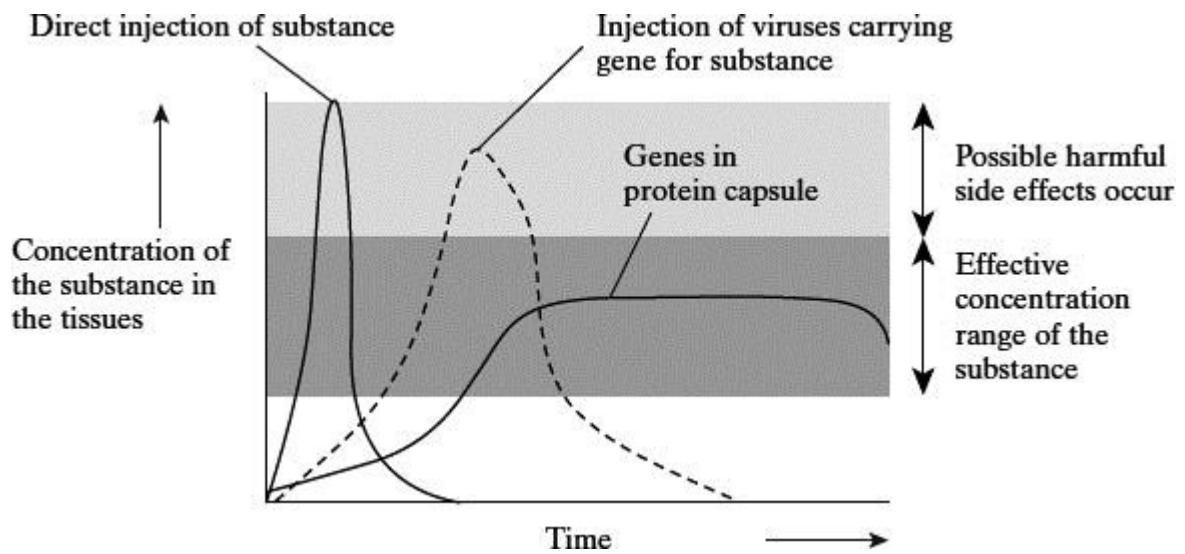
Q34.

- [illegible]

- (b) In gene therapy, genes are introduced into a person who has defective genes which do not produce an important substance. Three experiments were done to compare techniques for introducing an important substance into a person with defective genes.

1. The substance was injected directly.
2. Harmless viruses carrying genes coding for the substance were injected.
3. The genes were put into a protein capsule which was inserted into the tissues.

The graph shows results of the experiments.



Takahiro Ochiya et al, Biomaterials for Gene Delivery: Studies on Metastasis, (National Cancer Centre, Research Institute, Tokyo, Japan) 1999

- (i) Describe the results of the three experiments.

- (ii) Using the information in the graph, suggest **one** advantage and **one** disadvantage of the capsule method compared to the others.

Advantage _____

Disadvantage _____

Q35.

- (a) Describe how a gene can be isolated from human DNA.

(2)

- (b) Describe how an isolated gene can be replicated by the polymerase chain reaction (PCR).

(4)

- (c) (i) Describe how a harmless virus, genetically engineered to contain a CFTR gene, can be used to insert the gene into a cystic fibrosis sufferer.

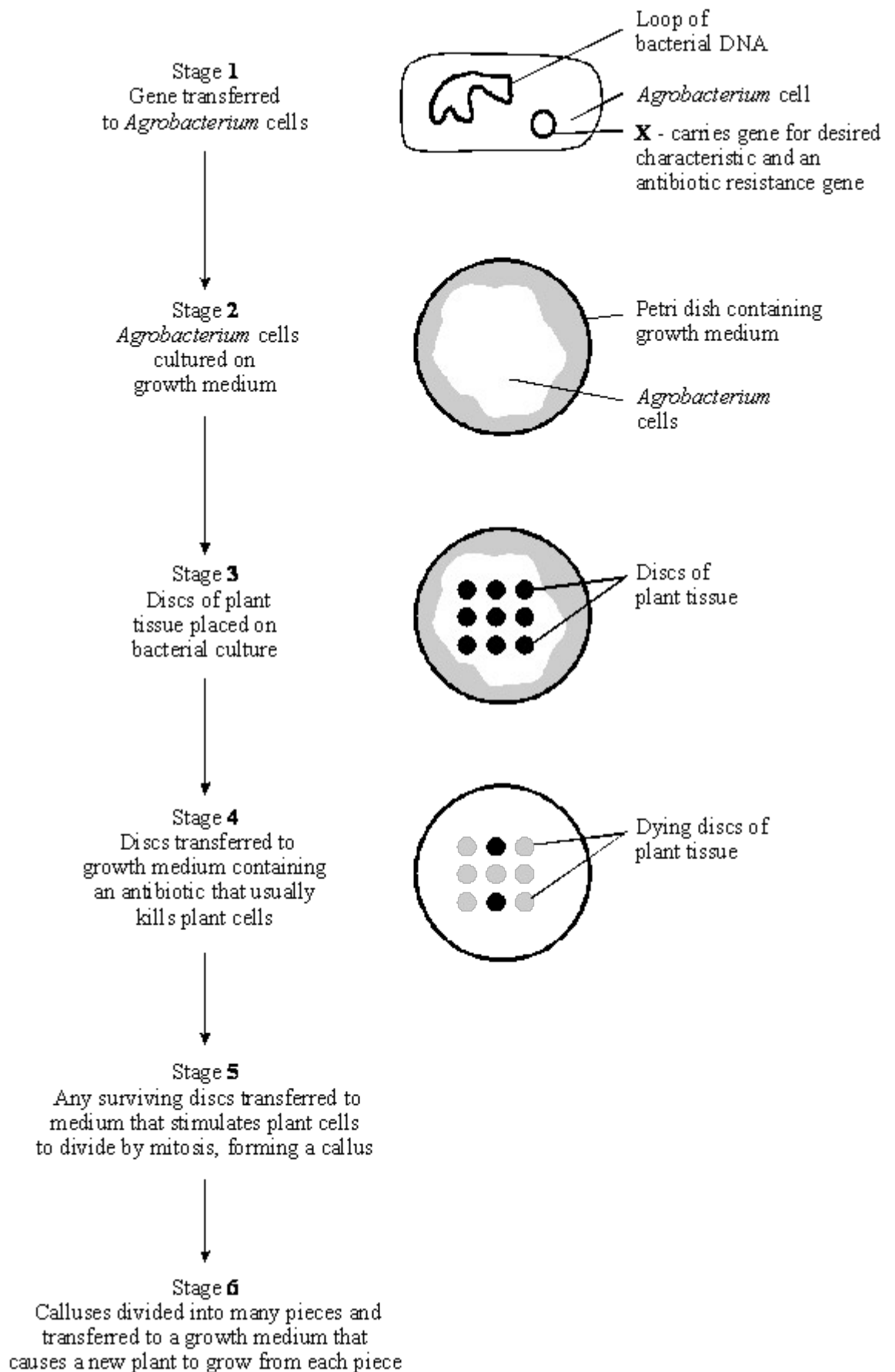
(2)

- (ii) A virus used in gene therapy has RNA as its genetic material and has an enzyme called reverse transcriptase. Inside a human cell, reverse transcriptase uses viral RNA to make viral DNA.

Explain why the enzyme is called *reverse transcriptase*.

Q36.

- (a) *Agrobacterium* is a bacterium used in genetic engineering of plants. The diagram shows stages in the transfer of a gene into a plant.



(i) Name structure **X** in stage 1.

(1)

- (ii) In stage **2**, explain why the bacteria are cultured before the plant tissue is added.

(1)

- (iii) In stage **4**, explain why the growth medium contains antibiotic.

(2)

- (iv) Suggest why stages **5** and **6** are necessary for the commercial production of genetically engineered plants.

(2)

- (b) (i) A toxin that kills insects can be sprayed directly onto the leaves of crop plants. A gene has now been transferred into crop plants that makes their leaves produce this toxin.

Explain **one** advantage to farmers of growing the genetically engineered crop plants, rather than spraying leaves with the toxin.

(1)

- (ii) Suggest **one** reason why some people are concerned that the toxin gene might get transferred to wild plants that are related to the crop plants.

(1)

(Total 8 marks)

Mark schemes

Q1.

- (a) (All) the DNA in a cell/organism;
Accept
 '(all) the 'genes'/alleles' 'genetic material/code' in a cell/organism/ person'
 'the total number of DNA bases in a cell/organism'
Reject all the DNA/ genes within a species
 1
- (b) 1. (Transcriptional factor/antibody) has a specific/tertiary structure/shape;
Accept (antibody) has a specific variable region
Accept (transcription factor/antibody) has a specific binding site
Reject active site but only once.
 2. Complementary (shape/structure);
Reject active site but only once.
 2
- (c) DNA, transcription factor and antibody;
Accept Nucleotides for DNA
Ignore 'reference to chemicals'
 1
- (d) Binds to P34 gene/DNA/mRNA
OR
 Binds to transcription factor gene/DNA
OR
 Binds to promoter;
Reject binds to transcription factor
 1
- (e) 1. Restriction (endonuclease/enzyme) to cut plasmid/vector;
 2. Ligase joins gene/DNA to plasmid/vector;
 2
- (f) 1. Mass/number of amino acids/polypeptides;
Accept weight for mass
Ignore density/size
Accept length of polypeptide/amino acid chain
Accept primary structure /sequence of amino acids.
Accept tertiary structure
 2. Charge;
 3. R groups (differ);

2 max

[9]

Q2.

- (a) 1. No (functional) cones
OR
Only rods;
2. Cones are connected to a single neurone
OR
Several rods connected to a single neurone;
Accept correct reference to retinal convergence
Accept 'bipolar/nerve cell' for neurone
Accept 'many' 2 or more for 'several'
3. (Cones) Separate (sets of) impulses to brain
OR
(Rods) Single (set of) impulse/s to brain;
Accept 'optic nerve' for brain
Reject 'signals', 'messages' for 'impulses'
Accept 'action potential'
- 3
- (b) 1. Correct answer in range 42 – 44% = 2 marks;;
2. Incorrect answer but shows that understanding that $2pq$ = heterozygous/carriers = 1 mark;
Accept $1 - (p^2 + q^2)$
Accept understanding of $2pq$ by using calculation involving 2 × two different numbers
- 2
- (c) 1. (Gene/allele) is on the X chromosome;
2. Females require two alleles/females can be heterozygous/carriers **and** males require one allele;
Reference to allele is essential but only required once
*Reference to females **and** males required*
Reject dominant allele
- 2
- (d) 1. Green sensitive pigment/cones non-functional
OR
Cones that detect green light non-functional;
2. Three different types of pigment/cone;
3. Other/different colours ('seen') due to stimulation of more than one cone/pigment;
1, 2 and 3. Reject reference to 'green cones'/'blue cones'/'red cones' but once only
1, 2 and 3. Reject reference to 'green pigment'/'blue pigment'/'red pigment but once only and only if 'green cones' etc, (see above) has not been rejected.
- 3
- (e) 1. (iPS cells) divide;

2. (iPS cells) develop/differentiate into (green sensitive) cones;
Accept 'produce'/'specialise' 'turn in to' / 'genes switched on' / 'turned on' for 'develop' but ignore 'grow'
Reject develop into 'green cones'/blue' cones'/'red cones'
Ignore develop/differentiate into (blue/red sensitive) cones;
Reject reference to develop in to 'green pigment'/'blue pigment' / 'red pigment'

2

- (f)
1. (Use of iPS cells) long-term;
Accept 'gene therapy short-term' or 'only two years'
Accept 'permanent'
 2. (Use of iPS cells) less chance of rejection/immune response;
 3. (Use of iPS cells) single treatment;
Accept 'gene therapy 'regular/frequent treatment''
 4. Harm/side effects from using viruses (in gene therapy);

3 max

[15]

Q3.

- (a)
1. Compete (with fertile males) to mate / for food / resources
OR
intraspecific competition;
 2. Do not reproduce / breed
OR
 Reduces population (of mosquitoes);
 1. *Must convey idea of competition.*
 2. *Accept: 'fewer mosquitoes' / 'fewer offspring'.*
- (b)
1. Capture / collect / sample, mark **and** release;
 2. Leave time for mosquitoes / Aedes to disperse before second sampling / collection;
 3. (Population =) number in first sample × number in second sample divided by number of marked in second sample / number recaptured;
 3. *Accept: correct equation.*
- (c)
- (Radiation) affects their 'attractiveness' / courtship / survival / life span;
*Accept: 'die / less likely to survive **due** to radiation'.*
Accept: 'disease can be transmitted by other means' (other than mosquitoes).
- (d)
- To maintain number / competition as they die / have a short life span;
Accept: to replace mosquitoes that have died.
- (e)
1. Number (of mosquitoes in treated area) is low / lower at / after 12/13/14/15/16 weeks = **2 marks**;

2

3

1

1

2. For one mark accept number (of mosquitoes in treated area) is low/lower without reference to relevant week;
Accept: amount for number.
Accept: comparison of numbers (of mosquitoes) for lower/low.

2

[9]

Q4.

- (a) Produces (c)DNA using (m)RNA;
Accept: 'converts' (m)RNA to (c)DNA.
Reject: tRNA
- (b) Joins nucleotides to produce (complementary strand/s of) DNA;
Accept: 'joins DNA nucleotides'.
- (c) 1. To remove any DNA present;
 2. As this DNA would be amplified / replicated;
 1. *Must be idea of removal / destruction.*
 2. *Accept: idea of DNA not being used as template.*
- (d) 1. Ratio in range of 1.4 :1 to 1.5 :1 = **2 marks**;
 2. One mark for answers which shows incorrect ratio but Shows 0.24 as a number or line on the graph
OR
 Ratio in correct range, but the wrong way round
OR
 Ratio in correct range but not expressed to 1
OR
 Ratio shown the other way round in range 1: 0.67 to 1:0.71;
Note: ratio not expressed to 1 in correct range may be shown in different ways, for example as: 3:2 or simply as 1.5 for one mark.
- (e) Limited number of primers / nucleotides;
Accept: DNA polymerase (eventually) denatures
Accept: primers / nucleotides 'used up'.
- (f) 1. Base sequences differ;
 2. (Different) complementary primers required;
 1. *Accept: reference to either RNA or DNA base sequences but reject reference to DNA base sequence in viruses.*

1

1

2

2

1

2

[9]

Q5.

- (a) 1. Human DNA / human gene / HGH gene contains introns
OR
 Methods 2 and 3 produce DNA / *HGH* without introns;
 2. *E. coli* cannot remove introns / cannot splice mRNA / cannot splice pre-mRNA;
 2
- (b) Faster to use gene machine than all the enzyme-catalysed reactions (involving reverse transcriptase);
Accept extra step / more steps involved in isolating mRNA
 1
- (c) 1. Cut the plasmid with a restriction endonuclease;
Allow 'add base sequences to blunt ends of plasmid and HGH gene'
 2. (So that) both have complementary / sticky ends;
 3. (Mix together) and add ligase to join the complementary / sticky ends;
 3
- (d) Can quickly identify transformed bacteria using UV light;
 1
- (e) 1. Arabinose alters structure of araC protein / reduces effect of araC protein;
 2. So stops / reduces inhibition of promoter gene and GFP gene is transcribed;
OR
 So stops / reduces inhibition of promoter gene and GFP is produced;
 2

[9]

Q6.

- (a) 1. (If injected into egg), gene gets into all / most of cells of silkworm;
 2. So gets into cells that make silk.
 2
- (b) 1. Not all eggs will successfully take up the plasmid;
 2. Silkworms that have taken up gene will glow.
 2
- (c) Promoter (region / gene).
 1
- (d) 1. So that protein can be harvested;
 2. Fibres in other cells might cause harm.
 2

[7]

Q7.

- (a) (i) Restriction endonuclease;
 1
- (ii) (DNA) ligase;
 1
- (b) (For those plants that contained the desired gene in the

nucleus/plant DNA)

1. (DNA of desired gene) copied/replicated with host DNA/inside nucleus;
2. Passed on by mitosis/plant grows by mitosis;
3. Produces genetically identical cells/clones;
Ignore references to protein synthesis or plasmids not taking up the gene
1. Accept DNA replication during mitosis
1. and 2. Accept converse for plants with the gene in the cytoplasm
3. Neutral 'identical unqualified'
3. Accept description, e.g., DNA is the same

3

- (c)
1. Genetic code is universal/triplets in DNA always code for same amino acid;
 2. It/insect DNA can be transcribed;
 3. Can be translated (process/mechanism same in all organisms/cells);
2. Accept (basic) transcription (process/mechanism) same in all organisms/cells;
2. Accept descriptions of process
3. Accept descriptions of process

3

[8]

Q8.

- (a)
1. Cut (DNA) at same (base) sequence / (recognition) sequence;
Accept: cut DNA at same place
 2. (So) get (fragments with gene) **R** / required gene.
Accept: 'allele' for 'gene' / same gene
- (b)
1. Each has / they have a specific base sequence;
 2. That is complementary (to allele r or R).
Accept description of 'complementary'
- (c)
1. Fragments L from parent rr, because all longer fragments / 195 base pair fragments;
Ignore: references to fragments that move further / less, require identification of longer / shorter or 195 / 135
Accept: (homozygous) recessive
 2. Fragments N from parent RR, because all shorter fragments / 135 base pair fragments;
1 and 2 Accept: A3 for 195 and A4 for 135
2. Accept: (homozygous) dominant
 3. (M from) offspring heterozygous / Rr / have both 195 and 135 base pair fragments.

2

2

Accept: have both bands / strips

Reject: primer longer / shorter

3

- (d) 1. (Cells in mitosis) chromosomes visible;
2. (So) can see which chromosome DNA probe attached to.

2

- (e) (i) 1. For comparison with resistant flies / other (two) experiments / groups;
Ignore: compare results / data / no other factors
2. To see death rate (in non-resistant) / to see effect of insecticide in non-resistant / normal flies.

Accept: 'pesticide' as 'insecticide'

Accept to see that insecticide worked / to see effect of enzyme

2

- (ii) (PM must be involved because)
1. Few resistant flies die (without inhibitor);
2. More inhibited flies die than resistant flies;
3. (PM) inhibited flies die faster (than resistant flies);
(Other factors must be involved because)
4. Some resistant flies die;
5. But (with inhibitor) still have greater resistance / die slower than non-resistant flies.

Accept: (with inhibitor) die slower than non-resistant flies

4 max

[15]

Q9.

- (a) Reverse transcriptase;

1

- (b) 1. Probe (base sequence) complementary (to DNA of allele A / where A is (and) binds by forming base pairs / hydrogen bonds;

Accept gene A

2. So (only) this DNA labelled / has green dye / gives out (green) light;

Accept glows for green light

2

- (c) (i) 1. More probe binding / more cDNA / mRNA / more allele / gene A means more light;
2. DNA (with **A**) doubles each (PCR) cycle;
3. So light (approximately) doubles / curve steepens more and more (each cycle) / curve goes up exponentially / increases even faster;

3

- (ii) (**G** because)

1. (Heterozygous) only has half the amount of probe for **A** attaching / only half the amount of DNA / allele A (to bind to);

Accept only one A to bind to

2. (So,) only produced (about) half the light / glow / intensity (of H)
(per cycle of PCR);
If reference to 'half' for point 1, allow 'less light' in 2.

2

[8]

Q10.

- (a) 1. No effect at 25°C
The question only refers to plants with GB
1. Reject same mass
2. Keeps growing at 30°C and 35°C / up to 35°C (more than without GB);
3. Above 35°C, falls but grows more than plant without GB;
3. Accept at all temperatures above 25°C more growth than without GB

2 max

- (b) (i) Significantly different / SEs do not overlap ;
Accept converse without GB

1

- (ii) (As temperature increases,)
1. Enzyme activity reduced / (some) enzymes denatured;
2. Less photosynthesis, so fewer sugars formed;
3. Less respiration / less energy / ATP for growth;
4. Less energy for named function associated with growth
4. Eg mitosis, uptake of mineral ions

4

- (c) 1. (Rubisco activase attaches to thylakoid and) this changes shape / tertiary structure (of enzyme) / blocks active site / changes active site;
Note - question states enzyme stops working when it attaches to thylakoid, not before
1. Accept rubisco in this context
2. (This) prevents substrate / RuBP entering active site / binding;
2. Accept prevents ES complex forming
2. Accept no longer complementary to substrate / RuBP

2

- (d) 1. GB prevents / reduces binding of rubiscoactivase to (thylakoid membrane);
1. Accept enzyme instead of rubiscoactivase. Accept rubisco
2. (Prevents it) up to 35°C;
3. (So) rubiscoactivase / enzyme remains active;

4. (So) photosynthesis / light-independent stage still happens;
4. Accept descriptions of light-independent stage
5. Above 35°C, some binding still occurs but less than without GB, so less reduction in growth;

4 max

- (e) 1. Looked for information / journals, on crop plants that grow at high temperatures;
1. "other research" is minimum accepted
1. Accept previous experiments research with temperature resistant crops
Ignore simple references to looking at previous studies / other plants - need to relate to this context
2. (Crop plants cited in this research) contain / make GB;
 3. So assumed making plants produce GB makes them resistant to high temperatures;

2 max

[15]

Q11.

- (a) Restriction / endonuclease;
Ignore specific names of restriction enzymes e.g. EcoR1
- (b) (i) 1. (Acts as a) marker gene to show that the (human) gene has been taken up / expressed;
1. Accept: gene marker
2. (Only) implant cells / embryos that show fluorescence / contain the jellyfish gene;
- (ii) 1. Factor IX present in / extracted from milk;
2. Gene only expressed in mammary glands / udder / gene not expressed elsewhere;
2. Ignore references to milk
The 'only' aspect is important here.
3. Do not need to kill sheep (to obtain Factor IX);
- (c) (i) 1. Mutation / nucleus / chromosomes / DNA may be damaged / disrupts genes;
1. Neutral: cell may be damaged
2. May interfere with proteins (produced) / gene expression / translation;
Ignore references to hormone levels or time of implantation

2 max

OR

3. Embryo / antigens foreign;
3. Neutral: antigens change
4. Embryo is rejected / attacked by immune system;
4. sNeed idea that the immune system is involved if mark point 3 has not been given
'Embryo foreign so rejected' = 2 marks
'Embryo rejected by immune system' = 1 mark
'Embryo is rejected' = 0 marks

2 max

- (ii) 1. Saves time / money for others;
2. Same work is not repeated / methods can be compared / improved / amended / same errors are not made;

2

[9]

Q12.

- (a) 1. Adenylate cyclase activated / cAMP produced / second messenger produced;
2. Activates enzyme(s) (in cell so) glycogenolysis / gluconeogenesis occurs / glycogenesis inhibited;
2. Neutral: 'glucose produced' as given in the question stem
Accept: correct descriptions of these terms

2

- (b) (i) 1. Glucose / sugar in food would affect the results;
1. Accept references to starch / carbohydrate
Or
2. Food / eating would affect blood glucose (level);
Or
3. (Allows time for) blood glucose (level) to return to normal;
3. Neutral: allows time for insulin to act

1 max

- (ii) Type 2 diabetes is a failure to respond to insulin / still produces insulin / is not insulin-dependent;

1

- (iii) (For) – 3 max

A maximum of three marks can be awarded for each side of the argument

1. Avoids injections / pain of injections;
2. Long(er) lasting / permanent / (new) cells will contain / express gene;
Ignore references to methodology e.g. sample size not known
3. Less need to measure blood sugar / avoids the highs and lows in

blood sugar;

4. Less restriction on diet;

(Against) – 3 max

5. Rats are different to humans;

6. May have side effects on humans;

6. *Accept: virus may be harmful / disrupt genes / cause cancer*

7. Long(er) term effects (of treatment) not known / may have caused effects after 8 months;

8. (Substitute) insulin may be rejected by the body;

4 max

[8]

Q13.

- (a) (i) 1. Negative correlation;
Accept: description for 'negative correlation'
Neutral: 'correlation'
Reject: positive correlation
2. Wide range;
3. Overlap;
4. (Graph suggests that) other factors may be involved (in age of onset);
2 / 3 Accept the use of figures from the graph
2 / 3 Can refer to age of onset or number of CAG repeats
Ignore references to methodology
- (ii) 1. Age of onset can be high / symptoms appear later in life;
Accept: 'gene' for 'allele'
2. (So) individuals have already had children / allele has been passed on;

3 max

OR

3. Individuals have passed on the allele / already had children;
4. Before symptoms occur;

2 max

- (b) (i) 1. Person **K**;
2. (As has) high(est) band / band that travelled a short(est) distance / (er) so has large(st) fragment / number of CAG repeats;
Must correctly link distance moved and fragment size

2

- (ii) Run fragments of known length / CAG repeats (at the same time);
Accept: references to a DNA ladder / DNA markers
Do not accept DNA sequencing
- (iii) Homozygous / (CAG) fragments are the same length / size / mass;
Accept: small fragment has run off gel / travelled further

1

1

[9]

Q14.

- (a) 1. Closer the (amino acid) sequence the closer the relationship;
2. (Protein structure) related to (DNA) base / triplet sequence;
Amino acid sequence is related to (DNA) base / triplet sequence = two marks;
- (b) 1. Reference to base triplets / triplet code / more bases than amino acids / longer base sequence than amino acid sequence;
Different (base) triplets code for same amino acids = 2 marks;
Degeneracy of triplet code = 2 marks
2. Introns / non-coding DNA / degeneracy of code / more than one code for each amino acid;
Ignore reference to codon.

2

2

[4]

Q15.

21 – 25	Extended abstract Generalised beyond specific context	Response shows holistic approach to the question with a fully integrated answer which makes clear links between several different topics and the theme of the question. Biology is detailed and comprehensive A-level content, uses appropriate terminology, and is very well written and always clearly explained. No significant errors or irrelevant material. For top marks in the band, the answer shows evidence of reading beyond specification requirements.
16 – 20	Relational Integrated into a whole	Response links several topics to the main theme of the question, to form a series of interrelated points which are clearly explained. Biology is fundamentally correct A-level content and contains some points which are detailed, though there may be some which are less well developed, with

		appropriate use of terminology. Perhaps one significant error and, or, one irrelevant topic which detracts from the overall quality of the answer.
11 – 15	Multistructural Several aspects covered but they are unrelated	Response mostly deals with suitable topics but they are not interrelated and links are not made to the theme of the question. Biology is usually correct A-level content, though it lacks detail. It is usually clearly explained and generally uses appropriate terminology. Some significant errors and, or, more than one irrelevant topic.
6 – 10	Unistructural Only one or few aspects covered	Response predominantly deals with only one or two topics that relate to the question. Biology presented shows some superficial A-level content that may be poorly explained, lacking in detail, or show limited use of appropriate terminology. May contain a number of significant errors and, or, irrelevant topics.
1 – 5	Unfocused	Response only indirectly addresses the theme of the question and merely presents a series of biological facts which are usually descriptive in nature or poorly explained and at times may be factually incorrect. Content and terminology is generally below A-level. May contain a large number of errors and, or, irrelevant topics.
0		Nothing of relevance or no response.

Commentary on terms and statements in the levels mark scheme

The levels mark scheme for the essay contains a number of words and statements that are open to different interpretations. This commentary defines the meanings of these words and statements in the context of marking the essay. Many words and statements are used in the descriptions of more than one level of response. The definitions of these remain the same throughout.

Levels mark scheme word/statement	Definition
Holistic	Synoptic, drawing from different topics (usually sections of the specification)
A fully integrated answer which makes clear links between several different topics and the theme of the question	All topics relate to the title and theme of the essay; for example, explaining the biological importance of a process.

	When considering, for example, the importance of a process, the explanation must be at A-level standard. ‘Several’ here is defined as at least four topic areas from the specification covered. This means some sentences, not just a word or two. It does not mean using many examples from one topic area.
Biology is detailed and comprehensive A-level content, uses appropriate terminology, and is very well written and always clearly explained.	Detailed and comprehensive A-level content is the specification content. Terminology is that used in the specification. Well written and clearly explained refers mainly to biological content and use of terminology. Prose, handwriting and spelling are secondary considerations. Phonetic spelling is accepted, unless examiners are instructed not to do so for particular words; for example, glucagon, glucose and glycogen.
No significant errors or irrelevant material.	A significant error is one which significantly detracts from the biological accuracy or correctness of a described example. This will usually involve more than one word. Irrelevant material is several lines (or more) that clearly fails to address the title, or the theme of the title.
For top marks in the band, the answer shows evidence of reading beyond specification requirements.	An example that is relevant to the title and is not required in the specification content. The example must be used at A-level standard.
Response mostly deals with suitable topics but they are not interrelated and links are not made to the theme of the question.	Not addressing the biological theme of the essay (e.g. importance) at <u>A-level standard</u> .

Please note that to obtain full credit, students must use information to show **the importance of Using DNA in science and technology**.

Topics

DNA and classification	
2.2	Structure of DNA

2.3	Differences in DNA lead to genetic diversity
2.9	Comparison of DNA base sequences
Genetic engineering and making useful substances	
2.5	Plasmids
5.8	The use of recombinant DNA to produce transformed organisms that benefit humans
Other uses of DNA	
2.5	Cell cycle and treatment of cancer
5.8	Gene therapy; Medical diagnosis and the treatment of human disease; The use of DNA probes to screen patients for clinically important genes.

In order to fully address the question and reach the highest mark bands students must also include at least four topics in their answer, to demonstrate a synoptic approach to the essay.

Students may be able to show the relevance of other topics from the specification.

Note, other topics from beyond the specification can be used, providing they relate to the title and contain factually correct material of at least an A-level standard. Credit should not be given for topics beyond the specification which are below A-level standard.

[25]

Q16.

- | | | |
|-----|--|---|
| (a) | restriction (enzyme) / endonuclease / named example; | 1 |
| (b) | unpaired bases / sticky ends / staggered;
complementary / explained; | 2 |
| (c) | <i>1 mark for each correct outcome</i>
plasmid with foreign DNA joined in ring;
ring with plasmid only; ring of foreign DNA only;
<i>ignore linear structures</i> | 3 |

[6]

Q17.

- | | | |
|-----|---|---|
| (a) | Restriction (enzyme / endonuclease); | 1 |
| (b) | Move towards anode / move because charged;

Different rates of movement related to charge / size; | 2 |

- | | | | |
|---------|--|-------|------------|
| (c) (i) | Piece of DNA;
Single stranded;
Complementary to / binds to known base sequence / gene; | max 2 | |
| (ii) | DNA invisible on gel / membrane;
Allows detection; | 2 | |
| | | | [7] |

Q18.

- | | | | |
|---------|--|-------|-------------|
| (a) (i) | protein / immunoglobulin;
specific to antigen;
idea of 'fit' / complementary <u>shape</u> ; | 2 max | |
| (ii) | 1. virus contains antigen;
2. virus engulfed by phagocyte / macrophage;
3. presents antigen to B-cell;
4. memory cells / B-cell becomes activated;
5. (divides to) form clones;
6. by mitosis;
7. plasma cells produce antibodies;
8. antibodies specific to antigen;
9. correct reference to T-cells / cytokines; | 6 max | |
| (b) | 1. antibody gene located using gene probe;
2. cut using restriction enzyme;
3. at specific base pairs;
4. leaving sticky ends / unpaired bases;
5. cut maize / DNA / vector using same restriction enzyme;
6. join using DNA ligase;
7. introduce vector into maize / crop / recombinant DNA into maize; | 4 max | |
| (c) | passive / person is not making own antibodies / antibodies not replaced;
memory cells not produced; | 2 | |
| (d) | fewer ethical difficulties / less risk of infection; | 1 | [15] |

Q19.

- | | | | |
|---------|--|---|------------|
| (a) (i) | Reverse transcriptase; | 1 | |
| (ii) | Idea that mRNA is present in large amounts in cell making
the protein / mRNA has been edited / does not contain
introns / mRNA codes for single protein; | 1 | |
| (b) | (Ligase) splices / joins two pieces of DNA / "sticky ends"; | 1 | [3] |

Q20.

- | | | | |
|-----|--|---|------------|
| (a) | Endonuclease / restriction enzyme; | 1 | |
| (b) | DNA made of base pairs;
Each base pair is same length / occupies same distance
along backbone; | 2 | |
| (c) | (i) Second blank box from left labelled 6; | 1 | |
| | (ii) Distance moved depends on length / number of base pairs /
second longest fragment / second shortest distance identified; | 1 | |
| (d) | 5; | 1 | |
| | | | [6] |

Q21.

- | | | | |
|-----|--|---|------------|
| (a) | Mother and father both heterozygotes / Tt / carriers;
Probability of thalassaemia 1/4 and female 1/2;
Probability of both 1/8; | 3 | |
| (b) | (i) Cut at same base sequence as same enzyme used;
Fragments are same length / size / have same charge; | 2 | |
| | (ii) Single base occurs many times;
Sequence of 20 unlikely to occur elsewhere;
<i>Allow one mark for establishing the principle where neither marking
point clearly made.</i> | 2 | |
| | | | [7] |

Q22.

- | | | | |
|-----|--|---|------------|
| (a) | (i) <u>Sticky ends</u> / description;
Reference to complementary base-pairing | 2 | |
| | (ii) Ligase; | 1 | |
| (b) | Carrier of DNA / gene; (<i>context of foreign DNA</i>)
Into cell / other organism / host; | 2 | |
| (c) | Act as marker gene;
Allows detection of cells containing plasmid / DNA; | 2 | |
| | | | [7] |

Q23.

- | | | |
|---------|---|-------|
| (a) (i) | Different genes / characteristics / features;
Reference to mutations;
Or
Base sequence determines protein;
Different species have different protein sequences; | max 2 |
| (ii) | Primer has different DNA sequence;
DNA specific / complementary base-pairing; | 2 |
| (iii) | Electrophoresis separates DNA;
(So they can be) identified by position on gel;
Smaller / shortest fragments travel furthest / quicker / or
reverse argument; | 3 |
| (b) | (<i>conventional</i>) Many lengths / all DNA / (<i>new</i>) one length;
Each rung is DNA of one / specific length; | 2 |
| (c) | 1 Heat DNA;
2 Breaks hydrogen bonds / separates strands;
3 Add primers;
4 Add nucleotides;
5 Cool;
6 (to allow) binding of nucleotides / primers;
7 <u>DNA</u> polymerase;
8 Role of (DNA) polymerase;
9 Repeat cycle many times; | max 6 |
- [15]**

Q24.

- | | | |
|---------|--|---|
| (a) (i) | <u>Sticky ends</u> / description;
Reference to complementary base-pairing | 2 |
| (ii) | Ligase; | 1 |
| (b) | Carrier of DNA / gene; (<i>context of foreign DNA</i>)
Into cell / other organism / host; | 2 |
| (c) | Act as marker gene;
Allows detection of cells containing plasmid / DNA; | 2 |
- [7]**

Q25.

- | | |
|-----|---|
| (a) | 1 (DNA altered by) mutation;
2 (mutation) changes base sequence;
3 of gene controlling cell growth / oncogene / that monitors cell division;
4 of tumour suppressor gene;
5 change protein structure / non-functional protein / protein not formed; |
|-----|---|

- 6 (tumour suppressor genes) produce proteins that inhibit cell division;
- 7 mitosis;
- 8 uncontrolled / rapid / abnormal (cell division);
- 9 malignant tumour;

max 6

- (b) cancer cells die / break open;
releasing DNA;
- (c) normal DNA and changed DNA have different sequences;
DNA only binds to complementary sequence;
- (d) fewer abnormal / cancerous cells / smaller tumours;
less cell damage / less spread / fewer locations to treat;
- (e) mRNA base sequence has changed;
gene / DNA structure is different / has mutated;
cancer gene active / tumour suppressor gene inactive;

2

2

2

3

[15]

Q26.

- (a) (i) contains genes / nucleotides / sections of DNA / artificial
DNA from two species / 2 types of organisms;
- (ii) carries gene / DNA (into the other organism / gene carrier);
- (iii) expose cells to the fungus;
non-resistant ones die, resistant ones survive;
OR identify by adding marker gene / gene probe / (qualified)
marker probe; description of positive result
e.g. radioactivity / fluorescence / complementary base pairing;
- (b) EITHER 1 cut desired gene (from DNA) of oat plant;
2 using restriction endonuclease / restriction enzyme;
OR 1 use mRNA from oat which will code for resistance;
2 and use reverse transcriptase to form desired DNA;
OR 1 make artificial DNA with correct sequence of bases;
2 using DNA polymerase;
3 cut plasmid open;
4 with (same) restriction endonuclease / restriction enzyme;
5 ref. sticky ends / unpaired bases attached;
6 use (DNA) ligase to join / ref. ligation;
7 return plasmid to (bacterial) cells;
8 use of Ca^{2+} / calcium salts / electric shock;
(if ref. to 'insulin' allow 5 max.)

1

1

2

max 6

[10]

Q27.

- | | | |
|-----|--|-------|
| (a) | Presence of resistant and non-resistant varieties / mutation produces resistant variety;
Resistant ones survive / non-resistant ones killed by treatment;
These will reproduce and produce more resistant parasites / pass on resistance allele; | 3 |
| (b) | Likelihood of being infected (by strain resistant to both drugs) is less;
$1/500 \times 1/500/1/250\,000$;
Drug has longer effective life; | max 2 |
| (c) | (i) As comparison / to show that nothing else in the treatment was responsible; | 1 |
| | (ii) Given injections of saline / injection without SPf66;
(otherwise) treated the same as experimental group; | 2 |
| (d) | (i) 100%; | 1 |
| | (ii) 10%; | 1 |
| (e) | (i) Different lengths of DNA have different base sequences / cut at specific sequence;
Results in different shape / different shape of active site;
Therefore (specific sequence) will only fit active site of enzyme; | 3 |
| | (ii) Recognition sites contain only AT pairs;
Which would occur very frequently; | 2 |
- [15]**

Q28.

- | | | |
|-----|--|-------|
| (a) | (i) transfer / carry genes from one organism to another / into bacteria / cells; | 1 |
| | (ii) cut open plasmid;
cut donor DNA, to remove gene / length of DNA;
cut donor DNA and plasmid with the <u>same</u> enzyme / enzyme that cuts at the <u>same</u> base sequence;
sticky ends / (overhanging) ends with, single strand / bases exposed;
association / attachment / pairing of complementary strand; | 2 max |
| | (iii) annealing / splicing / backbones joined / phosphodiester bonds; | 1 |
| (b) | (i) L and M; | 1 |
| | (ii) fragments 64 and 36(kilobases obtained) | 1 |

Q29.

- (a) 1 macrophages present antigens to B lymphocytes;
 2 antigen binds to / is complementary to receptors on lymphocyte;
 3 binds to a specific lymphocyte;
 4 lymphocytes become competent / sensitised;
 5 (B) lymphocytes reproduce by mitosis / (B) lymphocytes cloned;
 6 plasma cells secrete antibodies;

4 max

- (b) 1 restriction enzyme / endonuclease;
 2 to cut plasmid / to form sticky ends in plasmid;
 3 (use) ligase(to join) gene to plasmid;
 4 culture bacteria with (in medium containing) plasmids
 5 to allow uptake of plasmids / transformation;
 6 use of cold shock / chemical treatment (to enhance uptake) / heat shock;
(ignore bullets / electroporation / microinjection)

3 max

[7]

Q30.

- (a) (i) restriction (endonuclease) enzyme;
cuts DNA at specific / restriction points / after specific base sequence;

2

- (ii) PCR / polymerase chain reaction;

1

- (b) isolated cells divide by mitosis;
 can get many plants (producing toxin) / rapid production of
 (toxin producing) plants;
all cells (in the new plant / clone) will produce the toxin;

3

[6]

Q31.

- (a) probe will attach (to mutant allele);
 attaches to one DNA strand;
 as a result of complementary base pairing;
 radioactivity detected on film / X-ray / by autoradiography
 (if mutant allele present);

4

- (b) for
 gene is only active in mammary cells / only affects milk / easy to
 obtain product / product produced in large amounts / gene passed to
 offspring;

1

against

long term effects not known / qualified reference to animal exploitation
 e.g. use of embryos / effect of inserted gene on other sheep

tissues / genes;

1

[6]

Q32.

(a) introduction of healthy gene / 'replacement' of defective gene;

1

(b) can enter cells / infect cells / inject DNA into cells;
targets specific cells;
replicates (in cells);

2

(c) reproductive cells / gamete cells do not contain ADA allele / gene;

1

(d) (i) to 'prevent' rejection / immune response;

1

(ii) T lymphocytes have a limited life span / die off / do not reproduce;
bone marrow provides continual supply of T lymphocytes /
(ADA) gene enzyme;

2

[7]

Q33.

(a) (cut out gene using an) endonuclease / restriction enzyme;
reference to specificity / recognition site;
sticky ends;
use the same enzyme to cut;
plasmid / virus / potato DNA;
fixed by ligase;
method of introducing vector e.g. micropipette / virus injects DNA /
remove plant cell wall;

6 max

(b) different genes are expressed;
producing different enzymes / proteins;

2

[8]

Q34.

(a) isolate wanted gene / DNA from another organism / mRNA from cell / organism;
using restriction endonuclease / restriction enzyme / reverse transcriptase to get
DNA and produce sticky ends;
use ligase to join wanted gene to plasmid;
also include marker gene e.g. antibiotic resistance;
add plasmid to bacteria to grow (colonies) then (replica) plate onto medium where
the marker gene is expressed;
bacteria / colonies not killed have antibiotic resistance gene and (probably) the
wanted gene;

6

(b) (i) injection, rapid rise and fall;
virus, slower rise and longer in effective / harmful range;

capsule slowest rise, longest in effective / harmful range;
 injection and virus give harmful concentrations but capsule does not;

3 max

- (ii) advantage e.g.:
 substance never reaches harmful levels / no side effect / less likely to harm the organism, longer relief from symptoms / less frequent treatment needed / longer effective range / longer but without harmful side effects;

1 max

disadvantage e.g.:
 takes longer to take effect;

1

[11]

Q35.

- (a) use restriction enzyme / endonuclease / named, e.g. Bam / Eco;
 to cut DNA in specific place / base sequence;

2

- (b) heat DNA to 90 – 95 °C;
 strands separate;
 add primers;
 and nucleotides;
 cool so that primers bind to DNA;
 (DNA) polymerase forms new strands / joins nucleotides;

4 max

- (c) (i) virus is inhaled / sprayed into the lungs;
 gets into cells, inserting the healthy gene;

2

- (ii) makes DNA from RNA
rather than other way round

1

[9]

Q36.

- (a) (i) plasmid;

1

- (ii) the bacteria divide / grow, producing many copies of desired gene / plasmid;
 OR
 the bacteria divide / grow to cover the agar;

1

- (iii) plant tissue that has antibiotic resistance survives;
 identifies plant tissue which has desired gene / plasmid;

2

- (iv) to clone plants / produce genetically identical plants with gene / characteristic;
 and produce large numbers / quickly;

2

(b) (i) (*one reasonable suggestion*),
e.g. toxin present all the time;
save costs of buying / application of spray;
no spray drift onto other fields / insects;

1 max

(ii) (*one reasonable suggestion*),
e.g. killing of harmless / useful insects that feed on wild plants;
damage to food chains starting with wild plants;

1 max

[8]

Examiner reports

Q1.

- (a) The mark scheme for this question reflected the considerable variation in acceptable definitions for the term genome. Despite this, nearly 40% of students did not obtain the mark. There some very detailed definitions which often included the DNA in mitochondria and chloroplasts. The most common incorrect responses referred to “all the genes in a chromosome” or “all the genes in a species”.
- (b) Almost 80% of students obtained at least one mark for this question, often for referring to complementary structures. Almost half of these students gained a second mark by referring to a specific tertiary structure, shape or binding site. Weaker responses suggested that the term complementary is equivalent to ‘similar’ or referred to ‘active site’ rather than binding site.
- (c) This proved more difficult than expected with only 26% of students obtaining the mark. Incorrect responses included (unbound) DNA fragments, nucleotides, exons, introns, DNA without an antibody or transcription factor bound to it, and any combination of two of the three parts required to gain the mark. The constituent parts of a DNA molecule were also listed.
- (d) Again, relatively few students (30%) obtained the mark for this question. The most frequent correct responses referred to cDNA binding to the *P34* gene, preventing its transcription, or binding to mRNA, preventing its translation. The binding of the cDNA to mRNA, resulting in the destruction of mRNA, was also described. Binding to the promoter region was less frequently awarded. A significant minority of students mentioned that cDNA prevented RNA polymerase binding to the *P34* gene or just prevented its transcription, but with no suggestion of how. Incorrect responses often referred to cDNA increasing/decreasing methylation of the *P34* gene or simply that cDNA could not be transcribed into mRNA and translated to form the *P34* protein.
- (e) Considering that this question was relatively straightforward, it proved to be a very effective discriminator. Surprisingly, many students could not name both enzymes correctly. Consequently, only 35% of students obtained both marks for this question and 28% obtained a single mark. DNA helicase, reverse transcriptase and DNA polymerase were frequent incorrect responses. Even when the correct enzymes were named, students did not always describe the precise roles of the enzymes in the formation of recombinant plasmids. These descriptions were often too vague, e.g. “produces sticky ends” or “joins DNA together”, without any mention of the plasmid/vector. A significant minority referred to incorrect types of bonds when outlining the role of correctly named enzymes.
- (f) A significant number of students had difficulty applying their knowledge of electrophoresis to separating proteins. Only 14% of students gained both marks for this question, however 50% did at least obtain a single mark. Many students simply reverted to discussing DNA, especially VNTRs, or referred to DNA and protein, often suggesting that proteins consist of bases. Common responses which were not credited often referred to ‘size’ and ‘mass’, with no reference to the structure of proteins. However, better answers did obtain a mark when mass/size was linked to the number of amino acids or polypeptides. Difference in charge was the most frequently awarded marking point. Far fewer students mentioned different ‘R groups’ or ‘variable groups’.

Q2.

- (a) Despite only 8% of students obtaining maximum marks, this question proved to be an effective discriminator. 90% of students obtained at least one mark, often for stating that only rods or no (functional) cones are present. Slightly more than half of these students gained a second mark, usually for correctly describing the connections of rods and/or cones to a single neurone. However, poor terminology prevented some of these students from obtaining maximum marks. Some students used terms such as 'messages' and 'signals' rather than impulses. Even when students did refer to impulses, they often omitted reference to the optic nerve or brain, resulting in incomplete explanations.
- (b) The majority of students (70%) gained at least one mark for recognising that $2pq$ represented the heterozygous genotype. 39% of students successfully completed the calculation to obtain both marks. Many students scoring zero simply provided the Hardy-Weinberg equation with no annotation. Students obtaining one mark often thought that 0.1 was q rather than q^2 , resulting in $2pq = 2 \times 0.9 \times 0.1$. This mark was not awarded when students (commonly) suggested that pq represented the heterozygous genotype.
- (c) Almost 70% of students obtained at least one mark for this question, with both of the marking points being awarded in similar numbers. Students who scored zero often correctly stated that red-green colour blindness is sex-linked but did not mention the X chromosome in their explanation. These students often realised that males only required one recessive allele to express the condition, but did not explain that females require two alleles. Some students did refer to males and females, but omitted the term allele. Sex-linked alleles were sometimes said to be on an unspecified sex chromosome, on both X and Y, or just on the Y chromosome preventing females being affected. Females were sometimes thought to be XY and males XX.
- (d) The answers to this question were extremely disappointing. Less than 1% of students gained maximum marks and only about a third of students obtained any of the three marks available. The trichromatic theory is a new topic on the specification and many students displayed a poor understanding of how different colours are distinguished. Many students incorrectly referred to green/red/blue cones/pigments rather than green-/red-/blue-'sensitive' cones/pigments. Consequently, fewer than 5% gained a second mark. Although better students did appreciate that 'other colours' were mixtures of different wavelengths of light, they failed to explain that 'other colours' resulted in the stimulation of more than one type of cone at the same time. Students who did gain a mark invariably referred to non-functional green-sensitive pigments or cones. However, this point was often negated as many students suggested that red-sensitive cones were also non-functional or absent, or that all cones were non-functional or absent. It was not uncommon to read answers that provided details on the breakdown of pigments which were not only beyond the requirements of the specification, but were also often inaccurate.
- (e) Only 3% of students gained both marks in this question as it was a rarity to see responses that mentioned that iPS cells would divide. Although most students did understand that iPS cells would differentiate, many suggested they formed the 'green-sensitive pigment' rather than cone cells. A significant proportion of students referred to the iPS cells developing into green/red/blue cones, again failing to gain the mark for omitting the term 'sensitive'. There were also answers which referred to iPS 'growing' into cone cells with no indication of development or differentiation. Some students suggested that rod cells were produced or confused this method with gene therapy and referred to genes being injected into stem cells. Consequently, 48% of students scored zero on this question.

- (f) Most students (70%) obtained at least one mark for this question. Usually, this mark was awarded for stating that iPS is a long-term treatment or that gene therapy would only last two years. An additional mark was available if students realised that iPS would be a single treatment or that gene therapy would require regular treatments. However, this idea was rarely conveyed in students' answers. Far more students appreciated that one advantage of iPS was that rejection or an immune response was less likely. Although many students mentioned that gene therapy could cause 'harm' or 'side effects', this was often in the context of the 'injection' rather than due to using viruses. Consequently, only 8% of students obtained maximum marks and only 33% gained two or more marks. Many students also mentioned ethical issues, cost and/or the idea that results on monkeys were not necessarily transferable to humans. However, these constraints could also have applied to the use of iPS cells as a treatment.

Q3.

- (a) The idea of reducing the success of reproduction of the mosquito was well understood by the vast majority of students. Any reference to competition was far less often included, resulting in only approximately 10% of students gaining both marks. A significant number of students thought that being infertile prevented sterile males from passing on the virus.
- (b) This question also proved to be a very effective discriminator. Although over 50% of students obtained at least two marks, less than half of these students went on to gain full marks. The most common omission was not mentioning leaving sufficient time for the mosquitoes to disperse before second sampling. A significant minority also failed to refer to releasing the mosquitoes after first sampling. When provided, the equation for calculating the population size was often correct.
- (c) Very few students gained the mark for this question, with many responses suggesting that sterile mosquitoes could still transmit the virus, or that the overall population size was too large for the method to have an effect. The most common acceptable suggestions involved the radiation shortening the life span of the mosquitoes, or female mosquitoes not being attracted to infertile males.
- (d) Approximately a third of students gained this mark, usually by suggesting that releasing more transgenic males replaced those that had died. Unfortunately, most answers simply referred to mosquitoes dying without any reference to the need to maintain the number of transgenic mosquitoes in the population.
- (e) The vast majority of students gained at least one mark for stating that the number of mosquitoes was lower in the treated area. Over 60% of students gained a further mark for specifying when this reduction in number became significant.

Q4.

- (a) The role of reverse transcriptase in forming cDNA, using mRNA, was fairly well understood, with almost two-thirds of students obtaining the mark. Some responses suggested that the enzyme formed RNA from DNA. Others confused the role of the enzyme with that of other enzymes such as DNA polymerase, RNA polymerase, restriction endonucleases and DNA helicase. The enzyme was also linked to forming hydrogen bonds between complementary bases and the formation of double stranded DNA or RNA.
- (b) Slightly more than a third of students obtained the mark for this question. These students often provided detailed answers which explained the role of DNA polymerase, including the formation of phosphodiester bonds. Incorrect responses

often referred to DNA polymerase joining bases together, either by phosphodiester or hydrogen bonds. Many other students omitted any reference to either DNA or nucleotides; these incomplete descriptions were not credited.

- (c) It was clearly evident from the responses that most students misinterpreted what was required in this question, or simply did not know why the DNA was hydrolysed. Consequently, only 3% of students obtained both marks for this question, and almost 80% scored zero. A common misconception was that hydrolysis results in hydrogen bonds being broken to make the DNA single stranded, so that primers and / or nucleotides could bind, so that it could be replicated. Another frequent response was that hydrolysis of the DNA would provide an extra source of nucleotides for the Polymerase Chain Reaction. Some responses did suggest that the hydrolysis of DNA would ensure that only the RNA would then lead to amplification of DNA; the mark scheme required students to clearly state that DNA was removed or 'destroyed'.
- (d) Almost 8% of students did not attempt this question. Despite this, over 55% of students obtained at least one mark for showing some understanding of how to arrive at the correct answer. Students who scored one mark often failed to represent a ratio in the correct format. Common errors were to give the ratio the wrong way round – a ratio in the range from 1:0.67 to 1:0.71 was frequently seen – or not to express the ratio to 1; a frequent response here being 0.054:0.037. One mark was also awarded for incorrect answers which showed that the intensity of fluorescence at which the number of cycles reached 50% of the maximum was 0.24. Another problem leading to an incorrect ratio being calculated was inaccuracy in reading the number of cycles from the graph. These answers often gained one mark, as the intensity of fluorescence was usually correctly shown as 0.24. Despite the steps involved in calculating the correct ratio, almost one in four students obtained a correct answer.
- (e) Less than 50% of students gained this mark. The most common incorrect responses were to suggest that a stop codon had been reached, DNA helicase had become denatured, or that there were no more bases rather than no more nucleotides available.
- (f) Almost 60% of students obtained at least one mark, often for stating that the primers are 'complementary'. However, when referring to the RNA viruses, many students often omitted either the term 'base' or 'sequence', so that only one in five students gained both marks. A typical response was "the primers are complementary to different respiratory diseases". A minority of students incorrectly suggested that the viruses contained DNA sequences.

Q7.

This whole question discriminated well across the range of marks.

- (a) 78% of students named the correct enzyme in (i) and 70% in (ii).
- (b) This question caused problems for many students, with 54% failing to gain any marks. This was because they did not read the flow chart carefully, did not take at face value the information given and re-wrote the question. Good answers started with the observation that plants that did contain the desired gene in all their cells had the plasmid DNA inserted into plant DNA in the nucleus and that this meant the desired gene would be replicated (with the host DNA). They then went on to state that growth of the plant would involve mitosis and that this would produce genetically identical cells (with the desired gene). Many students speculated that not all the plasmids would contain the gene, for which there was no evidence. By far the

commonest error was to read the question as 'Explain why the desired gene is expressed in cells.' Students who made this error often wrote at length (often onto additional pages) about transcription and translation and gained no credit. Some students wrote about DNA replication by transcription.

- (c) This part produced the second greatest discrimination in the paper, just less than the essay. Just over 8% obtained all three marks. These students noted that the genetic code is universal for one mark, followed by stating that this meant the insect DNA could be transcribed and translated, for a second mark. 40% of students obtained two marks for just writing that the DNA could be transcribed and translated. Instead of the code being universal, some students wrote about DNA, or DNA bases, being universal and failed to obtain the mark. 33% failed to obtain any marks.

Q8.

- (a) Over half of students managed to communicate the idea that the same restriction enzymes cut at the same place / recognition sequence on DNA. Only just over 10 percent considered the context of the question and went on to say that this would give fragments containing the same gene (R).
- (b) About half of students obtained both marks in this part and over 40 percent obtained 1. Most managed to convey the idea that there was binding between complementary base sequences on a primer and DNA from an allele. The better answers conveyed the idea of a specific base sequence for each primer.
- (c) Some very good and clear answers were seen to this part and a third of students obtained 3 marks. These students identified the genotypes of L, M and N and explained how they identified them on the basis of the sizes of primer attached to each and how far the bands moved. Quite a large number went on at length about the offspring as represented by M but failed to identify L and N. A few thought that there were two types of offspring because there were two bands for M; they clearly did not understand the simple genetics of the cross.
- (d) This proved far more challenging than expected. The examiners were looking for the idea that chromosomes would be visible (as separate structures) and thus the scientists could see to which chromosome the probe was attached. Fewer than 10 percent got both marks.
- (e)
 - (i) There were quite a large number of attempts at generic 'How Science Works' answers to this part. 50 percent of students obtained 1 mark, usually for suggesting that the control was to see the effect of the insecticide or that it was for comparison with resistant flies. However, only around 10 percent gave both of these.
 - (ii) Most students focused on one aspect of the results and scored 2 marks. They frequently spotted that if the enzyme was the only factor in resistance, then the results for the controls and the resistant flies with the inhibitor would be the same. Others focused on evidence for the statement by comparing the resistant flies with resistant flies with the inhibitor added.

Q9.

- (a) About 80% of students identified the enzyme in this part as reverse transcriptase. The commonest wrong answer appeared to be restriction endonuclease.
- (b) Most students obtained one mark for stating that the DNA probe has a base sequence complementary to the DNA of allele A. A third obtained a second mark by

going on to state that this allowed it to bind to the target DNA by base pairing, or that this meant only target DNA gives off green light.

- (c) (i) About half of students obtained one mark in this part for noting that the more probe binding to allele A, the more green light there would be. Nearly a quarter obtained a second mark, usually for also noting that the light curve goes up exponentially (or described). Only a few, 14%, obtained all three marks. These students explained that this was because the amount of DNA doubles (approximately) with each PCR cycle.
- (ii) Answers to this part often made references to G being heterozygous because this person had *fewer* A alleles and thus *less* light was produced. The examiners were looking for more precise statements relating to half the amount of A with probe attached and half (approximately) the light produced (at any given time). A third of students obtained both marks.

Q10.

As a whole, this question tested students' understanding of the relationship between photosynthesis and the growth of plants. The questions were marked on outcome; this is to say that the examiners expected answers of A-level standard.

- (a) Many students failed to read the y axis carefully enough. All of the samples of plants increased in dry mass after 3 days, they all grew but some less than others. GB had no effect at 25°C, compared with plants without GB. Few students noted this and quite a number stated that GB produced more growth at all temperatures. Relatively few students made reference to the protection given to growth by GB up to 35°C. However, quite a few noted that growth was reduced less above 35°C with GB.
- (b) (i) There was only one mark available for this question and, with this in mind, students were required to refer to the standard error bars not overlapping, or to state that there was a significant difference between plants producing GB and those that weren't.
- (ii) Over a third of students obtained one mark, usually for linking a reduction in photosynthesis to a reduction in glucose (simple sugar) production. Some were also given credit for suggesting that the reduction could be linked to reduced enzyme activity. This was as far as most students went. Indeed, quite a large number wrote about reduced photosynthesis producing 'less food for the plant'. This was disappointing at A-level. For most students, their statement about reduced glucose production was simply followed by 'therefore growth falls'. There were very good answers that linked reduced glucose production to less respiratory substrate and thus less ATP / energy for growth. Others displayed understanding that sugars from photosynthesis form the basis for production of other organic substances and that these add to dry mass.
- (c) This was another question where some students failed to read the question carefully. A large majority correctly suggested that *when* the enzyme attaches to the thylakoid, this changes the shape of the enzyme, and / or its active site. They then went on to link this to a failure to bind to its substrate. Those who did not read carefully suggested that the enzyme was changed *before* binding to the thylakoid. This did not preclude them from scoring marks but made it less likely.
- (d) It was pleasing to see that the chain of evidence and logic was seen by most students. The number of marks they obtained tended to be a question of how much of the story they gave.

- (e) Many students ignored the statement in the stem that the hypothesis was developed on the basis of previous research. Instead, they reiterated the evidence from the study in the question. Good answers included the idea that research might have shown that crops in hot climates naturally produce GB.

Q11.

- (a) Most students correctly described the type of enzyme as 'restriction' or 'endonuclease'. The most common incorrect response seen was 'DNA polymerase'.
- (b)
 - (i) The students who gained full credit appreciated that the jellyfish gene acted as a marker allowing scientists to identify the cells that had taken up the human gene. However, very few students knew the significance of this in terms of only implanting embryos which fluoresced into surrogate sheep. It was not uncommon for some weaker students to state that the *DNA* would fluoresce or be visible under a microscope. A minority thought that the *milk* would glow green if the human gene had been taken up. There were also some students who confused this marker gene with using two different marker genes for antibiotic resistance. They thought that the human gene would disrupt the jellyfish gene. Hence, cells that had taken up the human gene would not glow. Unfortunately, poor expression let down some students. Typical examples included 'allow cells that have taken up the Factor IX protein to be identified', 'implant cells that glow into the *nucleus*' and 'the jellyfish gene acts as a *probe*'.
 - (ii) Very few students appreciated that the promoter allowed the human gene to be expressed only in the mammary glands, or that sheep would not have to be killed in order to recover Factor IX. Weaker students were, again, often let down by poor expression, such as 'the Factor IX gene will be in the milk'. Some simply repeated information given in the stem of the question regarding the promoter's role in allowing transcription. This was sometimes followed by a brief account of translation. Similarly, a minority of students thought that haemophilia can be treated by drinking milk containing Factor IX.
- (c)
 - (i) Both routes through the mark scheme were frequently seen in scripts from better students. Students who failed to score typically fell into one of two camps. Some thought that the embryos had failed to implant or had not been fertilised. Others thought that the hormone levels of the surrogate sheep were too low for successful implantation or that insufficient nutrients were available to support the growth of the embryo.
 - (ii) Most students scored one mark for the idea that reporting failed attempts would allow methods to be improved, or that the same work or errors would not be repeated. However, it was usually only the very best students who went on to explain that this would save time or money.

Q12.

- (a) This proved to be an excellent discriminator and just less than half of students gained full credit. This was usually for describing the production of cAMP and its effect on glycogenolysis or gluconeogenesis. Very few students were able to name the enzyme within the plasma membrane as adenylate cyclase. Similarly, there was sometimes confusion between the activation of this enzyme and intracellular enzymes. Some students thought that adenylate cyclase directly affects glycogenolysis or gluconeogenesis. This said, the correct use of scientific terms beginning with 'g' was generally good and only a minority of weaker students confused 'glycogen' and 'glucagon'. The terms 'glyconeogenesis' and 'glucogenolysis'

also appeared in weaker answers.

- (b) (i) Most students were aware that glucose in food would affect the results or that eating would affect the blood glucose concentration. Very few students referred to the importance of allowing the blood glucose concentration to return to normal.
- (ii) Students who failed to score typically stated that type 2 diabetes is not a genetic disease.
- (iii) The most common advantages given for gene therapy were the avoidance of injections and the longer-lasting effect. The most common disadvantages given were the possible side effects, which were usually linked to the virus, and that the long-term effects are not known. Surprisingly, relatively few students made reference to the fact that rats are different from humans. Weaker students seemed to rely on 'stock' *How Science Works* answers to gain credit for the 'against' side of the evaluation. They typically referred to unknown sample sizes and the influence of unnamed 'other factors'. These were not credited.

Q13.

- (a) (i) Although the negative correlation was usually indicated, only the better students appreciated the wide range in the age of onset or the overlap in values. Very few students were aware that the wide range in age of onset for the same number of CAG repeats suggests that other factors may also be involved.
- (ii) A minority of weaker students gave answers that were out of context. Some thought that the allele for Huntington's disease is recessive and would therefore be passed on to offspring without a person knowing. Others thought that people with Huntington's disease would survive well into adulthood and then reproduce.
- (b) (i) Most students correctly identified Person **K** as testing positive for Huntington's disease. They went on to explain that this person has the fragment that moved the shortest distance and linked this to a greater number of CAG repeats. A minority of students failed to link correctly the distance moved with the length of the fragment.
- (ii) Students who failed to gain credit often referred to using DNA sequencing or probes to highlight sequences and 'restriction mapping'.
- (iii) Misconceptions seen in responses by weaker students included partial digestion of DNA, Person **L** only having one allele of this gene and the probe not being able to attach to the other fragment.

Q14.

- (a) Over 40% of students failed to score on this question. Many of these students suggested that proteins consist of bases and the confusion between bases and amino acids pervaded their responses. Although a number of students did correctly refer to the sequence of amino acids, only better students linked the similarity of the amino acid sequence with a close evolutionary relationship between different species.
- (b) This question also proved challenging with less than 50% of students gaining any

marks. However, a significant number of students did gain one mark for reference to the triplet code and students appreciating the degeneracy of this code were able to gain both marks. Some students gained credit by referring to introns or non-coding DNA.

Q15.

Using DNA in science and technology

The very best essays from candidates who selected this option were outstanding. They reviewed, often in great detail, the relevant aspects of the specification although not always incorporating the role of DNA in the classification of organisms. Considering that much of the content of this essay could be drawn from this unit, it was surprising how poor many answers were. Understanding of techniques was often extremely limited, particularly *in vivo* gene cloning and the use of markers. Many essays presented no more than a broad overview either emphasising ethical issues at the expense of biological detail or failing to distinguish established practice from wishful thinking.

Q16.

Unit 2

- (a) This part was well answered by most candidates. Enzyme E was correctly named by almost all candidates, although a few 'restrictive' enzymes were seen.
- (b) This part was also well answered, and sticky ends were well known. A few weak candidates thought that sticky ends are complementary because they have the same shape rather than possessing complementary base sequences.
- (c) It was unusual for candidates to score full marks in this section. Some did not realise that the foreign DNA could join up or that the plasmid could re-form without any DNA being inserted. A common problem was that candidates offered more than three responses.

Unit 3

- (a) Most could name restriction enzymes but reverse transcriptase was a common error.
- (b) A weakness in expression often denied candidates access to both marks, but it was generally appreciated that complementary base pairing would be possible between the exposed sticky ends.
- (c) The request for drawings presented most candidates with a novel situation and tested their ability to apply understanding and follow their previous explanation. Given the concept that complementary sticky ends can join, it should have been possible to deduce that the plasmid ends could rejoin, the ends of the foreign DNA could join up or, the situation most were familiar with, the plasmid and foreign DNA could join together.

Q17.

- (a) Most identified the enzymes as restriction enzymes though the variety of spellings used meant that credit could not always be awarded.
- (b) This question showed the the idea of larger fragments moving slower or not as far was well understood although there was much incorrect biology relating to charge.

- (c) In (i) the idea of complementarity was understood and communicated well by many candidates although the fact the DNA probe is single stranded was not seen in many responses. In (ii) the majority of candidates obtained one of the available marks for the idea of detecting the location. Surprisingly few candidates stated that without a radioactive probe the location would not be visible. Many candidates had knowledge of fluorescent markers and gave a response based on this knowledge rather than answering the question. A common misconception was that radioactivity shows up when X-rayed.

Q18.

- (a) Few candidates scored two in the first part and vague references to 'fighting infections' were common as was confusion of antigens with antibiotics. In part (ii), some candidates appeared to know the sequence of events but in many cases limited marks were gained because of incorrect biology and discussion of previous infections which 'the body remembers and is ready to fight off the second time'. B and T cells were confused, the exact role of mitosis was not well known and few explained the specific antibody synthesis by plasma cells. Some answers used bacteria as the infective agent.
- (b) This section scored the majority of the marks for the question in most scripts. Problems arose mainly where candidates failed to apply their knowledge to the situation. As a result, the gene was transferred to a plasmid which was put into maize or left in a test tube. In a few cases, the first part of the question led to suggestions of infecting the maize with various blood cells.
- (c) Several good answers referring to passive immunity were seen here but many failed to gain marks through lack of precision in their answer.
- (d) Examiners were looking for more than a simple response about cost here. Valid ideas on the ethics of this work or the possibility of transferring a disease were awarded marks.

Q19.

Unit 2

- (a) Reverse transcriptase was usually given, but the incorrect DNA polymerase or endonuclease figured frequently. The explanation in (ii) was elusive with many thinking the mRNA was more accessible as it was in the cytoplasm or in single strands.
- (b) Answers ranged here from the insufficient 'ligation' to lengthy but incorrect descriptions of joining DNA by means of hydrogen bonds on the base pairs. Those who considered more carefully gave a correct answer.

Unit 3

The enzyme in (a)(i) was well known, though a few simply referred to transcriptase. In (a)(ii), it was common to read that mRNA is easier to obtain because it is in the cytoplasm rather than the nucleus. However, a fair number gave a valid explanation, such as the idea that introns have been removed, or that the gene will only code for the protein required. In (b), there was some confusion with restriction enzymes. Some thought the ligase would cut the gene. Others saw it as a kind of filler, filling the gap between the sticky ends, rather than forming bonds.

Q20.

Unit 2

- (a) Most had no trouble here in identifying endonuclease.
- (b) This was badly done since few mentioned or implied that DNA was made of base pairs and that these occupied the same distance along the backbone. Many made it obvious that they meant the distance from one chain to the other or went on to write about the distances in the electrophoresis gel.
- (c) Although the correct box was often located, the second from the right was almost as commonly given. Appropriate justification for the choice was rewarded in (ii) although many candidates suggested fragment 6 was the fifth rather than the second largest.
- (d) All figures were given here with 5 only rarely chosen, 6 being most common.

Unit 3

In part (a), restriction enzyme was correctly identified by almost all candidates. However, part (b) attracted very poor answers. Few commented that DNA is composed of base pairs, or that all base pairs have the same length. Many simply re-stated the question. In (c) (i), most candidates correctly identified the box, but a significant number identified the penultimate box. These candidates explained in (c)(ii) that the *longest* fragments moved furthest. Although they had the correct idea that DNA separates according to length, they had the relationship the wrong way round. In (d), there were few correct answers. Most gave 6.

Q21.

- (a) Figure 2 showed that the male involved in this cross possessed two alleles and this should have alerted candidates to the fact that the gene concerned was not sex-linked. Most of those who identified it as being autosomal, were able to explain that the probability of the next child having thalassaemia was one in four. Rather less success was enjoyed, however, when it came to combining probabilities, and the mathematics of multiplying fractions was evidently beyond the ability of a number of candidates.
- (b) Part (i) was generally answered well by those candidates who distinguished between the processes of electrophoresis, chromatography and centrifugation. The basis of separation in this case is the difference in charge and, in DNA, this translates into differences in length. The numerous references to such features as solubility suggested that the distinction between these three processes was not always secure. The difficulties encountered by many in part (ii) stemmed largely from confusion between the terms allele, base and probe. There was, as a result, much inaccurate biology and little opportunity for awarding credit.

Q22.

Part (a) (i) was well answered by about half the candidates. Weaker candidates simply repeated material from the stem, using the term 'staggered cuts' rather than 'sticky ends', and omitted details about complementary base-pairing. In (a) (ii), almost everybody gained the mark for ligase, though there were references to polymerase, reverse transcriptase and even lipase. In (b), there was considerable confusion about the term 'vector'. The commonest error was to describe the recipient bacterial cell as a vector. A few answered in terms of the mosquito which transmits malaria. While examiners found many excellent answers in (c) from better candidates, it is clear that many candidates

believed that plasmids alone could be cultured on agar plates and 'killed' by antibiotics.

Q23.

This question proved to be very demanding for many candidates, although better candidates were able to score all, or almost all, of the available marks. In (a) (i), many candidates recognised that different species of shark have different characteristics, but mostly failed to link these characteristics to proteins or to genes. In (a) (ii), some candidates did realise that each primer had a specific base sequence which would bind to the shark DNA by complementary base-pairing, but weaker candidates simply re-stated the information in the stem. In (a) (iii), many candidates limited their answers to stating that the DNA fragment had to be a different length in each species so it could be identified. How this enabled identification was not stated. On the other hand, good candidates wrote succinctly about electrophoresis separating DNA by size, and explained how the different length fragments would move different distances up the gel. Part (b) was poorly done by most candidates. It was common to find descriptions of genetic fingerprinting here. A few very weak students confused the 'rungs' with base pairs in the DNA molecule. Part (c) was the most accessible part of the question. A large proportion of candidates clearly knew the polymerase chain reaction and gave excellent accounts, often gaining full marks. A minority of very weak candidates confused PCR and transcription.

Q24.

- (a) Sticky ends were mentioned often but few candidates could describe them if the actual words were not used. The way sticky ends were used was badly explained. A large proportion of candidates suggested ligase as the enzyme but several improbable names were also mentioned.
- (b) Many candidates explained this quite well. The main pitfall was for those who discussed vectors of disease despite the clear indication that this was not required.
- (c) A common misconception appeared here, namely plasmids growing on agar, surviving and passing on their antibiotic resistance. It is gratifying that some candidates who recognized that the plasmids must be placed into bacteria went on to score both marks. Several thought putting antibiotic resistance in at this point would help to combat disease at some later stage.

Q25.

- (a) Many candidates gave a good account of the changes a mutation could produce and those with clear expression achieved full marks; many scored three or four marks. Uncontrolled cell division and malignant tumors were frequently referred to and some appreciated that genes which controlled cell division could have changed. References to benign tumours or cell mutations were irrelevant in the context of this question.
- (b) Very few candidates achieved marks here, mainly because they did not read the question. Whole cells in the blood were not required, but the understanding that cancer cells could burst or die and release their DNA was.
- (c) Few seemed to understand this and restated the question without reference to the changed base sequences to which the strip would bind.
- (d) This was generally well known. The main reason for failing to gain marks was a reference to an undefined 'it' which would be growing, dividing or spreading, causing undefined damage.

- (e) Here too some candidates who understood the problem found it hard to explain that changes in the mRNA would reflect mutations in the DNA and would show that a cancer gene was active.

Q26.

- (a) Only the most able could explain that two species were required here for recombinant DNA although many could describe a vector. The third part elicited some thoughtful responses along the lines of exposing the cells to the fungus. Less able candidates wrote about exposure to the disease, and most went into antibiotic resistance without applying it to the situation here.
- (b) Many candidates were able to gain maximum credit here in a very few lines of script. However, there were still the misconceptions that genes are cut out of the plasmid and that this is 'splicing'. Some failed to address the context, or discussed fermenters.

Q27.

- (a) Many candidates understood the basic principles of natural selection underlying this part of the question and better answers related these to the development of resistance in malarial parasites. Responses, however, were frequently marred by imprecise use of terms. Thus malarial parasites were variously described as developing resistance, immunity or, in some cases, allergies to the drugs concerned, while resistance was described as taking place in bacteria, the disease or even in the human population.
- (b) Evidence from BYA5 suggests that many candidates understand that probabilities are combined by multiplication. However, they were unable to apply this principle to the example in this part of the question. The most frequent response was to add the two figures. The resulting value of 1/250 then proved difficult to explain, and the simple idea that the probability of being infected by a strain of malarial parasite resistant to both drugs was much lower eluded most.
- (c) The concept of a control proved surprisingly unfamiliar to most candidates and even the best seldom progressed beyond explaining that a control offered a standard against which to compare the effectiveness of the vaccine. This idea should have given rise, in part (ii), to injection with saline only in an otherwise identically treated control group. Answers ranged from those who clearly failed to appreciate the nature of a control and discussed issues which were largely ethical in nature, to responses which were in varying degrees incomplete. Such responses included making sure that both groups "lived in the same place" or "were the same age", ignoring the fact that these were only part of a whole range of factors which should have been kept constant. Evidence from this question and from the coursework suggests that the issue of controls is one that needs to be addressed by centres.
- (d) Better candidates experienced little apparent difficulty in identifying the correct percentages here. Incorrect answers fell into no set pattern and most responses which could conceivably be given arose at least once.
- (e) Many candidates were obviously of the opinion that restriction enzymes function in a way that is totally different from other enzymes, and attempted to explain their specificity in part (i) in terms of base pairing. Others clearly understood the principles involved but neglected to relate their understanding of enzyme action to this particular question. A lack of precision characterised many of the answers to part (ii). Thus there were frequent references to adenine and thymine but not to these bases forming the restriction sites. However, most candidates were able to

equate the frequency of cutting to the small size of the resulting fragments.

Q28.

There were some excellent answers to this question but very few maximum marks, as many candidates failed to describe the role of DNA ligase in sufficient detail.

- (a) The roles of vectors and restriction endonucleases were well known and described clearly by the majority of candidates. As with the polymerase enzymes, many candidates seem under the impression that DNA ligase catalyses complementary base pairing. The majority of candidates failed to mention the joining of the backbones or used imprecise terminology, which was too ambiguous to gain the mark.
- (b) The majority of candidates interpreted the information given to gain both marks.

Q29.

Most candidates gained at least four marks in this question.

- (a) The majority of answers lacked detail and a clear understanding of the correct sequence of events. Candidates had to select the facts needed to answer the question. Many gained a mark for replication by mitosis/cloning. All the other marking points were seen, with stronger candidates gaining all the marks. Few mentioned the importance of specific B lymphocytes, or differentiation resulting in plasma cells that release antibodies.

Antigen, antibody and receptor were often confused. Many mentioned memory cells and T cells.

Many candidates gained more than half the marks because there were six points on the mark scheme.

- (b) A large number of students wasted time describing how to isolate the gene. The gene had already been isolated. Marks were gained for references to restriction endonuclease, plasmid and ligase. Few suggested how to transfer plasmid into bacteria.

Q30.

This proved to be a discriminating question with only the very best candidates gaining maximum marks.

- (a)
 - (i) The enzyme was correctly named by the substantial majority of candidates. Surprisingly, many then didn't gain the second mark, either by failing to mention the cutting of the DNA or by missing the specificity of the place of action.
 - (ii) A specification term, well answered.

- (b) There were very few answers worthy of credit. Most candidates could not apply the principles of mitosis to this question. The better candidates recognised the limited transfer of genes to cells within a whole plant but then could not link the cloning of individual cells to rapid production of many plants made of cells, all potentially able to express the gene.

Q31.

Not surprisingly, this question produced a wide range of marks. There were some excellent answers but even the most able candidates had difficulty obtaining maximum marks in part (a).

The vast majority of candidates obtained one or two of the four marks available. The first mark gained was by indicating that the gene probe would attach to the mutant allele. Some of these candidates obtained a second mark by stating that autoradiography could be used to detect radioactivity. However, a number of candidates suggested that 'firing X-rays' at the sample would suffice. Another misconception was that the individual radioactive nucleotides would line up with the mutant allele rather than a single radioactive strand. Very few candidates appreciated that the gene probe would attach to one DNA strand and even fewer candidates explained that this would occur as a result of complementary base pairing, despite the cues given in the stem of the question.

Only better candidates used their knowledge of the process involved to explain one argument for the use of genetically engineered sheep to produce alpha-1-antitrypsin. Generally these candidates referred to the product being in the milk or being produced in large quantities. Many candidates simply stated that you could treat cystic fibrosis, which was stated in the stem of the question. Fewer candidates gained a mark for explaining one argument against the use of these sheep. It was disappointing to see a large number of candidates simply referring to 'not playing God'. The most common correct response was to state that long-term effects are not known. Some candidates did refer to the low success rate of using sheep embryos in this process.

Q32.

Although only a few candidates obtained maximum marks on this question, most were able to gain between two and four marks.

- (a) Most candidates correctly described what is meant by gene therapy, although many weaker candidates referred to the 'removal of defective genes' or provided a definition of genetic engineering.
- (b) Most candidates obtained one mark for stating that a virus can enter a cell or inject DNA into a cell. A minority of candidates gained a second mark by referring to viral replication or to viruses targeting specific cells.
- (c) Approximately half the candidates correctly explained that the gametes of treated individuals do not possess the ADA allele and consequently they cannot pass it on to their children.
- (d) Most candidates gained the mark in part (i) by suggesting that using a genetically matched donor would help prevent rejection. Part (ii) proved to be more discriminating. Better candidates appreciated that T lymphocytes would have a limited life span and gained one mark. However, very few candidates gained both marks by explaining that transplanted bone marrow provides a continual supply of T lymphocytes with the ADA allele.

Q33.

Generally this question was well answered with most candidates obtaining at least four marks. However, part (b) proved difficult for the majority.

- (a) Answers to this question provided the full range of marks. Better candidates gave a detailed description of genetic engineering and had little difficulty in obtaining five of the six marks available. A significant number obtained maximum marks. Weaker candidates referred to the use of a restriction endonuclease and the production of 'sticky ends'. Many of these candidates also referred to the use of a plasmid but, unfortunately, not always in the correct context. There was also some confusion over the use of the enzyme ligase. Many candidates failed to realise that the gene was inserted into a plant cell but did not take the cell wall into consideration when describing the method of transfer. Nevertheless, the quality of answers to this question was pleasing.
- (b) This proved to be an effective discriminator, with only better candidates suggesting how different cells could be produced by different genes being expressed. The best candidates then linked this to the production of different enzymes or proteins. Many candidates referred to the use of hormones without displaying any understanding of how these may initiate gene expression.

Q34.

- (a) There were many good answers to this factual recall question and marks of 5 and 6 were common. Some candidates were confused between plasmids and bacteria and wrote about culturing plasmids on agar plates.
- (b)
 - (i) Many candidates displayed a rather poor ability to describe the results of the three experiments. Many candidates did obtain one mark for reference to the fact that the protein capsule method never produced harmful levels of the substance. Most candidates ignored the relative speed of rise into harmful concentrations between the direct injection and virus injection method. They also ignored the relative speeds of fall in concentration of the substance with the three methods.
 - (ii) Many candidates were able to suggest an advantage and a disadvantage of the capsule method. Some failed to gain marks because of the vague ways in which they expressed their answers. For example, 'It doesn't give very high levels' for an advantage, with no reference to concentrations with possible harmful side effects.

Q35.

This question gave many candidates the opportunity to display their knowledge of factual material from the specification. Candidates who had not learnt the material performed very badly.

- (a) Most candidates obtained one mark for reference to restriction enzymes. Many got a second mark for the idea that such enzymes target specific base sequences. Some were not awarded this second mark, because they stated that enzymes could be used which just cut out the required gene.
- (b) Many candidates scored two or three marks here, usually for references to heating to about 90 °C and adding primers and free nucleotides. Clear, coherent and correct answers about the whole process were relatively uncommon. There frequently seemed to be some confusion about why heating to 90 °C was carried out and why cooling to about 60 °C was necessary. It appeared that many candidates knew what

went into PCR but did not have a clear appreciation of how it works.

- (c) (i) Many candidates obtained one mark for the idea that the virus is inhaled into the lungs. Some then got the second mark for the idea that the virus carries the CFTR gene into target cells. Quite a few candidates failed to get these marks, because they introduced plasmids into their answer and tried to explain how they would be put into the virus, or how they would be inhaled or injected. This was despite the question stating that the virus had been genetically engineered and contained the required gene.
- (ii) Most candidates correctly explained that in this case mRNA was used in the production of DNA.

Q36.

This question produced a very wide spread of marks. Candidates frequently failed to gain marks for poor use of language and terminology. The question also caused problems for candidates who did not read the information given carefully. As an example, Stage 1 shows that a desired gene has already been introduced into *Agrobacterium* using a plasmid, so culturing of the bacterium produces bacteria with the gene.

- (a) (i) Most candidates correctly described X as a plasmid.
- (ii) The specification refers to plasmids being used to incorporate genes into bacterial cells. It also refers to rapid reproduction of such bacteria then producing many copies of these genes. Relatively few candidates linked culturing of the bacteria to the idea of producing many with the already introduced gene for a desired characteristic. Credit was given to candidates who suggested that the reason was to cover the growth medium with bacteria, to ensure infection of discs of plant tissue.
- (iii) Many candidates got confused here and wrote about identifying bacteria with antibiotic resistance, rather than plant tissue that now had the gene for resistance. Those who did get this mark often failed to get a second mark, because they failed to note that such tissue was also likely to have the desired gene, which is linked to the marker antibiotic resistance gene.
- (b) (i) Most candidates obtained this mark. Some candidates failed to score because of vague, non-specific answers, usually along the lines of, 'It's cheaper'. The examiners expected some explanation of why it might be cheaper.
- (ii) As with part (i), many candidates obtained this mark but others did not because of vague answers such as, 'It is dangerous to the environment'.

Q1.

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.

- 5 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.
- 10 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.
- 15 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.

- 20 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.

- (a) Assuming no one with AD died in 2014, calculate the annual percentage increase in AD cases in America for 2014 (lines 2–4).

Answer = _____ %

(2)

- (b) Explain how donepezil could improve communication between nerve cells (lines 7–9).

(3)

- (c) Suggest and explain **two** reasons why there is a high frequency of the E280A mutation in Yaramul (lines 13–15).

1. _____

2. _____

(2)

- (d) Explain why natural selection has **not** reduced the frequency of the E280A mutation in the population (lines 16–17).

(2)

- (e) 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)

- (f) One scientific study which analysed chromosome 14 involved 102 individuals. The scientists recorded a sample size of 204. In this sample they detected 75 E280A mutations but only 74 potential AD cases (lines 19–21).

Suggest explanations for the figures the scientists recorded.

(2)

- (g) Suggest why a DNA probe for the mutated triplet was **not** considered a suitable method for detection of the E280A mutation (lines 22–23).

(2)

(Total 15 marks)

Q2.

Mycobacterium tuberculosis causes tuberculosis. The DNA of *M. tuberculosis* contains a direct repeat (DR) region. The DR region consists of 43 different, non-coding base sequences called spacers. Each spacer is found in a specific place in the DR region. In different strains of *M. tuberculosis*, some of these spacers have been lost.

- (a) (i) The DR region consists of non-coding base sequences.

What is meant by a non-coding base sequence?

(1)

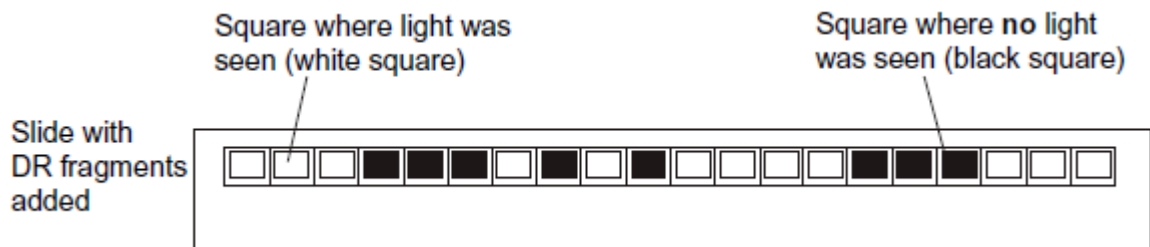
- (ii) Name the process by which the base sequence of a spacer is lost from a DR region.

(1)

Scientists investigated the DR regions of different strains of *M. tuberculosis*. They produced a DNA probe for each of the 43 spacer sequences. Each probe was:

- labelled with a fluorescent marker that gave off light if the probe attached to its complementary spacer
- attached to a particular square on a slide.

They obtained samples of the DR region from each strain. These were cut into small single-stranded DNA fragments. The fragments from each strain were added to a slide with the DNA probes attached. The diagram below shows their results for one strain of *M. tuberculosis* with 20 of the probes.



- (b) The scientists cloned the DR region DNA *in vitro* before testing for the presence of spacers.

Give the name of the method they used to clone the DNA *in vitro*.

_____ (1)

- (c) Explain how the use of DNA probes produced the results in the diagram.

 _____ (3)

- (d) Doctors can use the method with DNA probes to identify the specific strain of *M. tuberculosis* infecting a patient. This is very important when there is an outbreak of a number of cases of tuberculosis in a city.

Suggest and explain why it is important to be able to identify the specific strain of *M. tuberculosis* infecting a patient.

 _____ (2)

(Total 8 marks)

Q3.

Some populations of flies are becoming resistant to insecticides intended to kill them.

Scientists developed a method for finding out whether a fly was carrying a recessive allele, **r**, that gives resistance to an insecticide. The dominant allele, **R**, of this gene does not give resistance.

The scientists:

- crossed flies with genotype **RR** with flies with genotype **rr**
- obtained DNA samples from the parents and offspring
- used the same restriction endonuclease enzymes on each sample, to obtain DNA fragments.

- (a) Explain why the scientists used the same restriction endonuclease enzymes on each DNA sample.

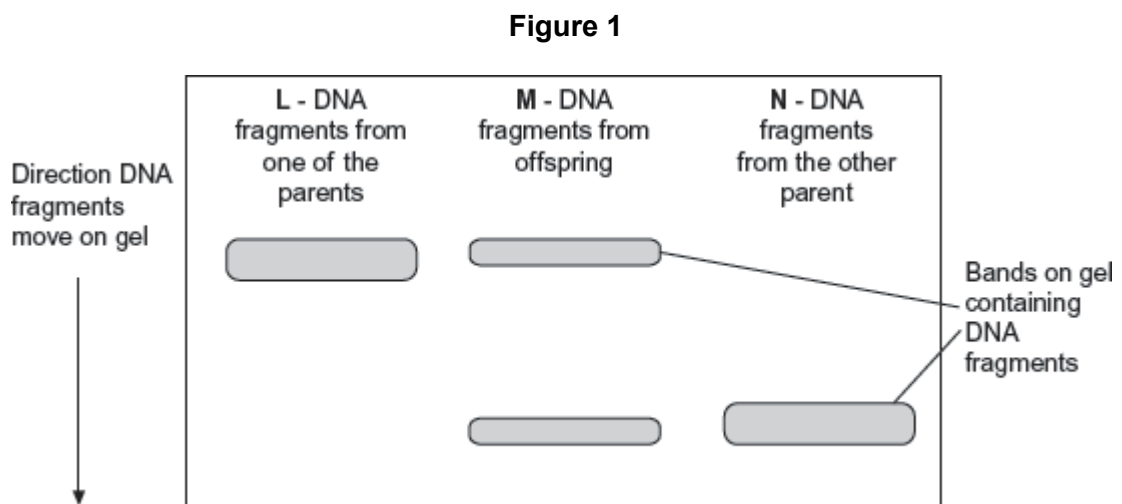
(2)

The scientists added two different primers to each sample of DNA fragments for the polymerase chain reaction (PCR).

- Primer A3 only binds to a 195 base-pair fragment from allele **r**.
- Primer A4 only binds to a 135 base-pair fragment from allele **R**.

The scientists separated the DNA fragments produced by the PCR on a gel where shorter fragments move further in a given time.

Their results are shown in **Figure 1**.



- (b) Explain why primer A3 and primer A4 only bind to specific DNA fragments.

(2)

- (c) Use all the information given to explain the results in **Figure 1**.

(3)

- (d) The scientists wanted to know on which chromosome the gene with alleles **R** and **r** was located. From the flies with genotype **RR**, they obtained cells that were in mitosis and added a labelled DNA probe specific for allele **R**. They then looked at the cells under an optical microscope.

Explain why they used cells that were in mitosis.

(2)

- (e) Another group of scientists thought that pesticide resistance in some flies was related to increased activity of an enzyme called P450 monooxygenase (PM). This enzyme breaks down insecticides.

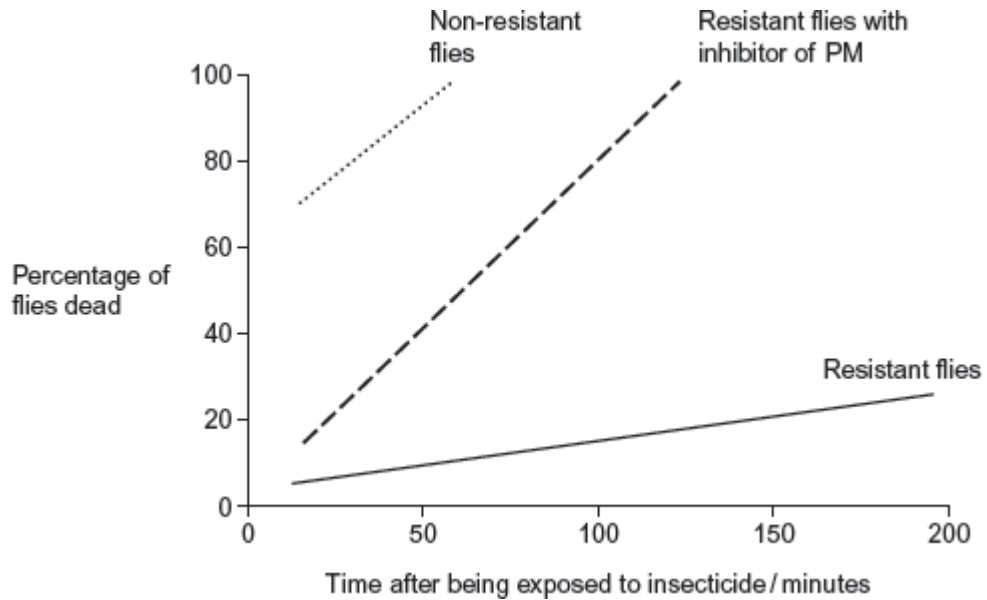
The scientists obtained large numbers of resistant and non-resistant flies. They then set up the following experiments.

- Non-resistant flies exposed to insecticide.
- Resistant flies exposed to insecticide.
- Resistant flies treated with an inhibitor of PM and then exposed to insecticide.

They then determined the percentage of flies that were dead at different times after being exposed to insecticide.

Figure 2 shows their results.

Figure 2



- (i) Explain why the scientists carried out the control experiment with the non-resistant flies.

(2)

- (ii) The scientists concluded that the resistance of the flies to the insecticide is partly due to increased activity of PM but other factors are also involved.

Explain how these data support this conclusion.

(4)

(Total 15 marks)

Q4.

Scientists wanted to measure how much mRNA was transcribed from allele **A** of a gene in a sample of cells. This gene exists in two forms, **A** and **a**.

The scientists isolated mRNA from the cells. They added an enzyme to mRNA to produce

cDNA.

- (a) Name the type of enzyme used to produce the cDNA.

(1)

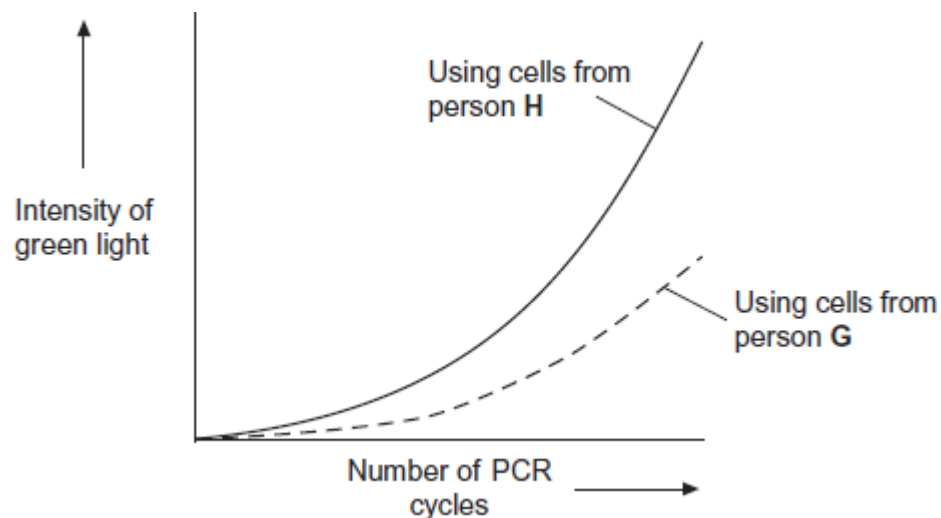
The scientists used the polymerase chain reaction (PCR) to produce copies of the cDNA. They added a DNA probe for allele **A** to the cDNA copies. This DNA probe had a dye attached to it. This dye glows with a green light **only** when the DNA probe is attached to its target cDNA.

- (b) Explain why this DNA probe will only detect allele **A**.

(2)

- (c) The scientists used this method with cells from two people, **H** and **G**. One person was homozygous, **AA**, and the other was heterozygous, **Aa**. The scientists used the PCR and the DNA probe specific for allele **A** on the cDNA from both people.

The figure shows the scientists' results.



- (i) Explain the curve for person **H**.

(3)

- (ii) Which person, **H** or **G**, was heterozygous, **Aa**? Explain your answer.

(2)

(Total 8 marks)

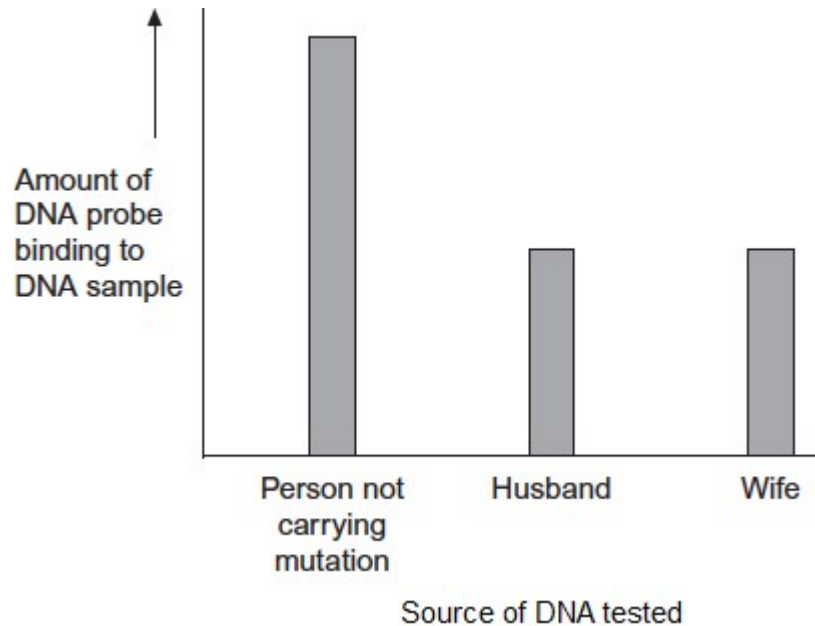
Q5.

A husband and wife wanted to know whether they were carriers of the mutated form of a gene. This mutation is a deletion that causes a serious inherited genetic disorder in people who are homozygous.

A geneticist took samples of DNA from the husband and the wife. He used a DNA probe to look for the deletion mutation. The DNA probe was specific to a particular base sequence in an exon in the gene. Exons are the coding sequences in a gene.

The geneticist compared the couple's DNA with that of a person known not to carry this mutation.

The chart shows the geneticist's results.



- (a) The geneticist told the couple they were both carriers of the mutated gene. Explain how he reached this conclusion.

(3)

- (b) The DNA probe the geneticist used was for an exon in the DNA, **not** an intron. Explain why.

(3)

- (c) To make the DNA probe, the geneticist had to find the base sequence of the normal gene. Once he had copies of the gene, what methods would he use to find the base sequence of the gene?

(2)

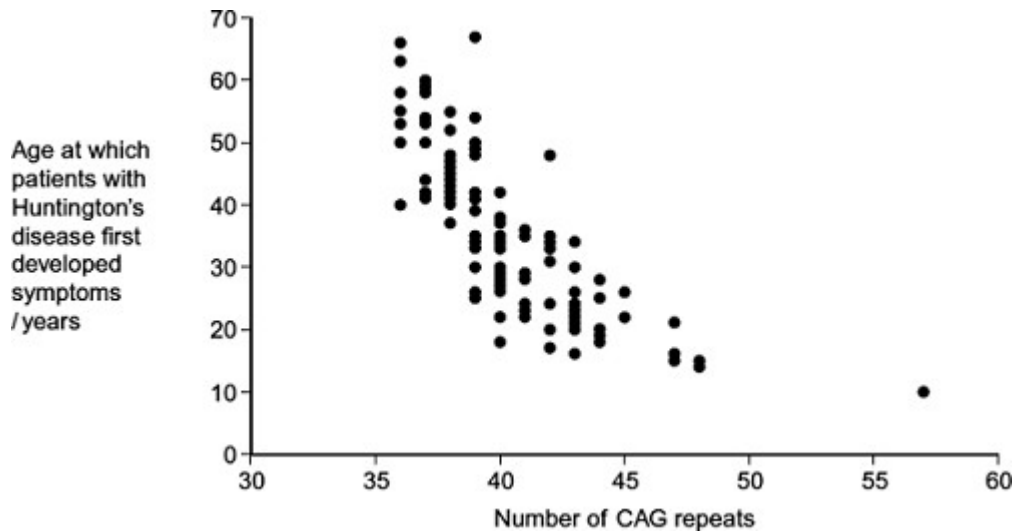
(Total 8 marks)

Q6.

Huntington's disease is a genetic condition that leads to a loss in brain function. The gene involved contains a section of DNA with many repeats of the base sequence CAG. The number of these repeats determines whether or not an allele of this gene will cause Huntington's disease.

- An allele with 40 or more CAG repeats will cause Huntington's disease.
- An allele with 36 – 39 CAG repeats may cause Huntington's disease.
- An allele with fewer than 36 CAG repeats will not cause Huntington's disease.

The graph shows the age at which a sample of patients with Huntington's disease first developed symptoms and the number of CAG repeats in the allele causing Huntington's disease in each patient.



- (a) (i) People can be tested to see whether they have an allele for this gene with more than 36 CAG repeats. Some doctors suggest that the results can be used to predict the age at which someone will develop Huntington's disease.

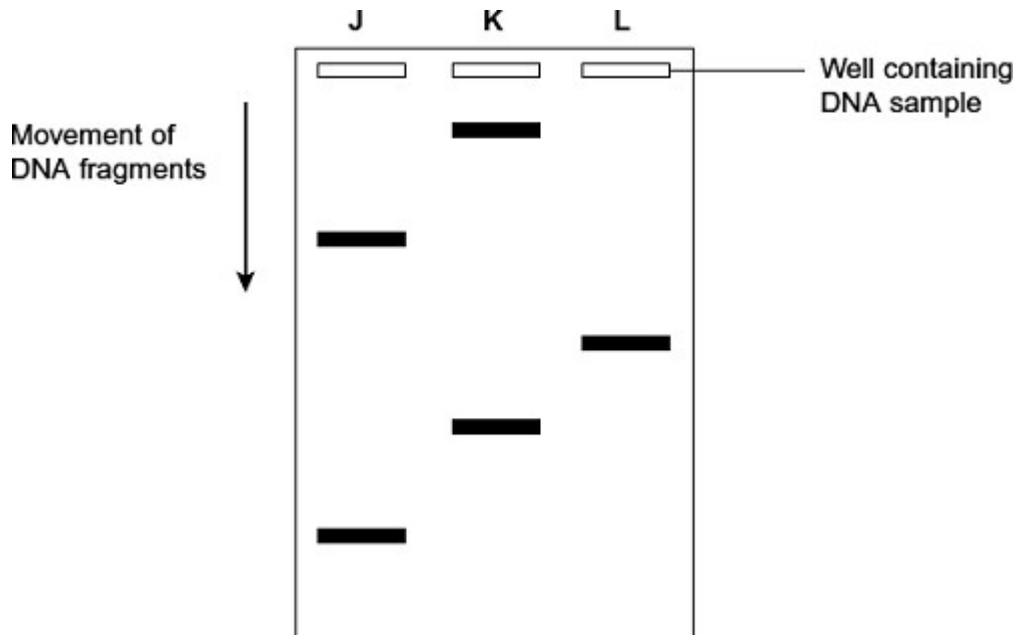
Use information in the graph to evaluate this suggestion.

(3)

- (ii) Huntington's disease is always fatal. Despite this, the allele is passed on in human populations. Use information in the graph to suggest why.

(2)

- (b) Scientists took DNA samples from three people, **J**, **K** and **L**. They used the polymerase chain reaction (PCR) to produce many copies of the piece of DNA containing the CAG repeats obtained from each person. They separated the DNA fragments by gel electrophoresis. A radioactively labelled probe was then used to detect the fragments. The diagram shows the appearance of part of the gel after an X-ray was taken. The bands show the DNA fragments that contain the CAG repeats.



- (i) Only one of these people tested positive for Huntington's disease. Which person was this? Explain your answer.

Person _____

Explanation _____

(2)

- (ii) The diagram only shows part of the gel. Suggest how the scientists found the number of CAG repeats in the bands shown on the gel.

(1)

- (iii) Two bands are usually seen for each person tested. Suggest why only one band was seen for Person L.

(1)

(Total 9 marks)

Q7.

- (a) Scientists can use protein structure to investigate the evolutionary relationships between different species. Explain why.

(2)

- (b) Comparing the base sequence of genes provides more evolutionary information than comparing the structure of proteins. Explain why.

(2)

(Total 4 marks)

Q8.

Write an essay on using DNA in science and technology.

(Total 25 marks)

Q9.

There are wolves in many European countries. Scientists investigated the genetic diversity of these wolves. They collected samples of DNA from the mitochondria of wolves from different countries. For each sample they identified which haplotypes were present in the DNA. A haplotype is a particular sequence of bases on DNA. Mutations can produce new haplotypes.

Country	Number of wolves sampled	Number of different haplotypes in mitochondrial DNA
Spain	84	3
Portugal	19	2
Italy	101	1
France	7	1
Bulgaria	29	6
Sweden	93	1

The scientists wanted to find out whether one of the haplotypes in the Portuguese wolves was the same as one of those in the Spanish wolves. They used a restriction endonuclease, electrophoresis and a labelled DNA probe.

(a) For what purpose did they use

(i) the restriction endonuclease

(1)

(ii) electrophoresis?

(1)

(b) Explain why the labelled DNA probe could be used to find out whether the haplotypes were the same.

(2)

(c) The scientists analysed the DNA on the Y chromosome and the DNA in the mitochondria of the Swedish wolves. They concluded that the Swedish wolf population descended from one male wolf from Finland and one female wolf from Russia.

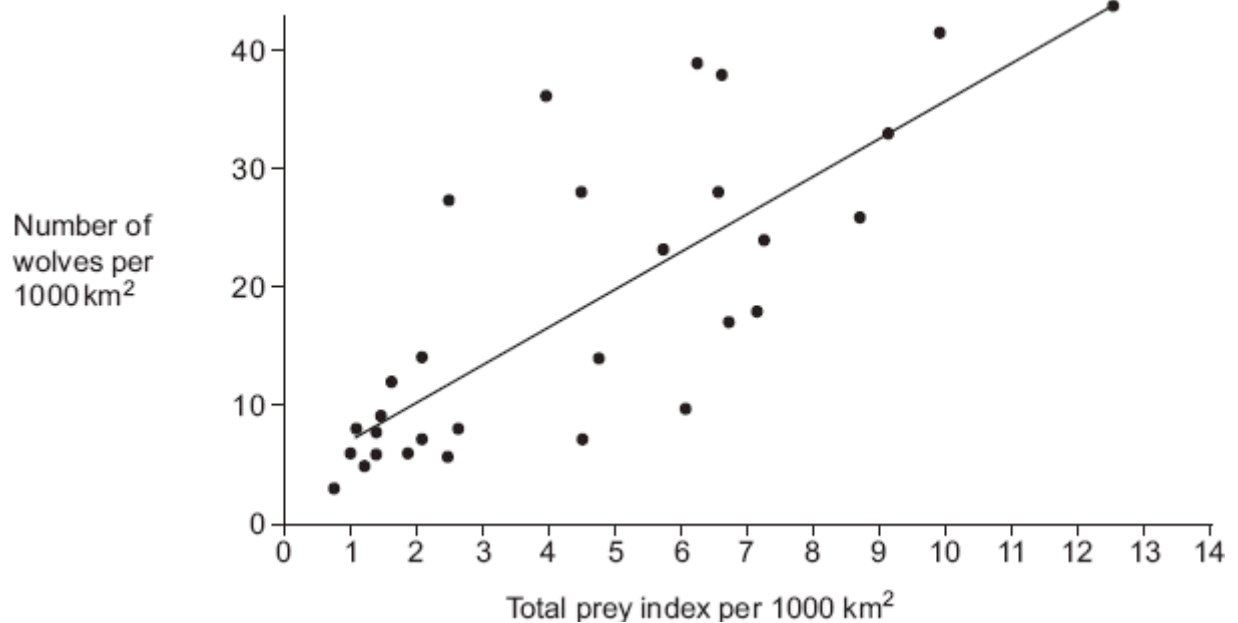
(i) Explain why DNA on the Y chromosome helped them to reach this conclusion.

(1)

(ii) Suggest why DNA in the mitochondria helped them to reach this conclusion.

(1)

Wolves eat different mammals. An ecologist investigated factors that affect wolf numbers in North America. He collected data from different field studies carried out in different places. The graph shows his results.



- (d) (i) The wolf numbers are given per unit area. Explain why.

(2)

- (ii) The ecologist calculated the total prey index for each of the places that had been studied. In order to do this, he gave each prey species a value based on how much food was available to wolves from the prey animal concerned. He called this value the prey index.

The ecologist considered that the prey index gave a better idea of the food available than the prey biomass in kg. Suggest why the prey index gives a better idea of food available.

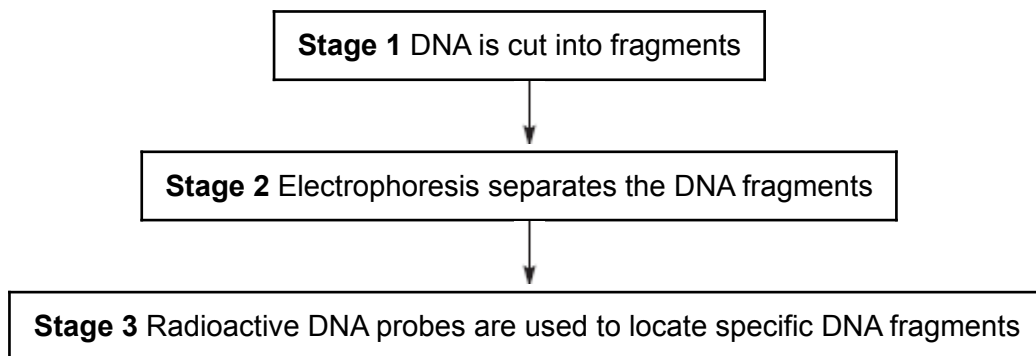
(2)

- (e) The ecologist calculated the total prey index by combining the prey indices and the total number of animals of each species present in 1000 km². He plotted this information on the graph. What does the graph suggest about the factors that determine wolf numbers in North America? Explain your answer.

(2)
(Total 12 marks)

Q10.

DNA probes may be used to identify the presence of specific genes associated with human diseases. The flow chart summarises the way in which they are used.



- (a) Name the enzyme used in **Stage 1**.

(1)

- (b) Explain how electrophoresis separates the fragments of DNA in **Stage 2**.

(2)

- (c) (i) What is a *DNA probe*?

(2)

- (ii) Explain why *radioactive* DNA probes are used to locate specific DNA fragments.

Q11.

A gene was broken into fragments using enzyme **Z**. The mixture of fragments produced was then separated by electrophoresis.

- (a) What type of enzyme is enzyme **Z**?

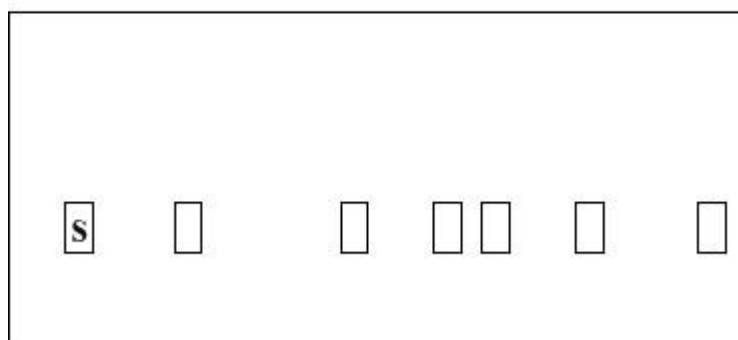
(1)

The table shows the number of base pairs present in the fragments.

Fragment	Number of base pairs ($\times 10^3$)
1	4.65
2	5.72
3	10.71
4	2.39
5	5.35
6	7.53

The diagram shows the electrophoresis gel used. The mixture of fragments was placed at the start point marked **S** and the process started. The boxes indicate the positions reached by the different fragments.

Direction of movement of fragments



- (b) Explain why base pairs are a suitable way of measuring the length of a piece of DNA.

- (c) (i) Write **6** above the appropriate box on the diagram to show the position you would expect fragment **6** to have reached.

(2)

(1)

- (ii) Explain how you arrived at your answer.

(1)

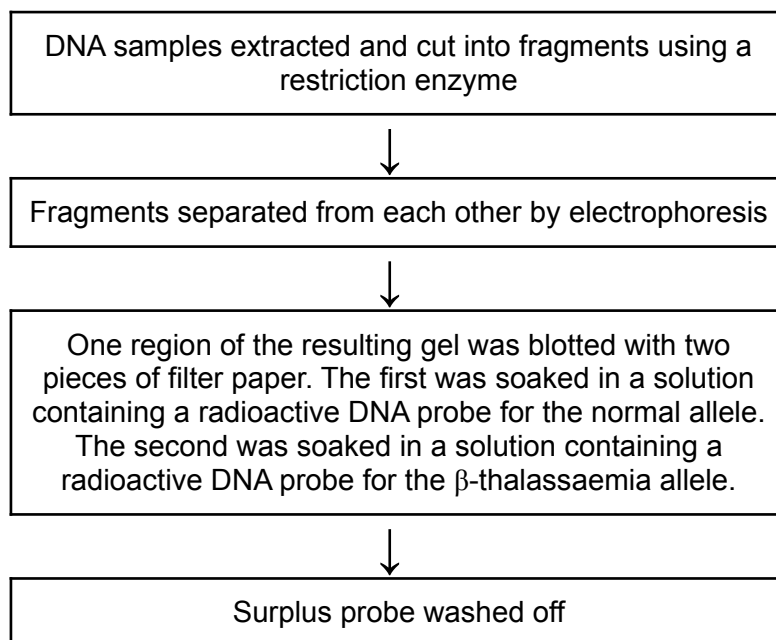
- (d) Enzyme **Z** recognises a particular sequence of bases in the gene. How many times does this sequence appear in the DNA of this gene?

(1)

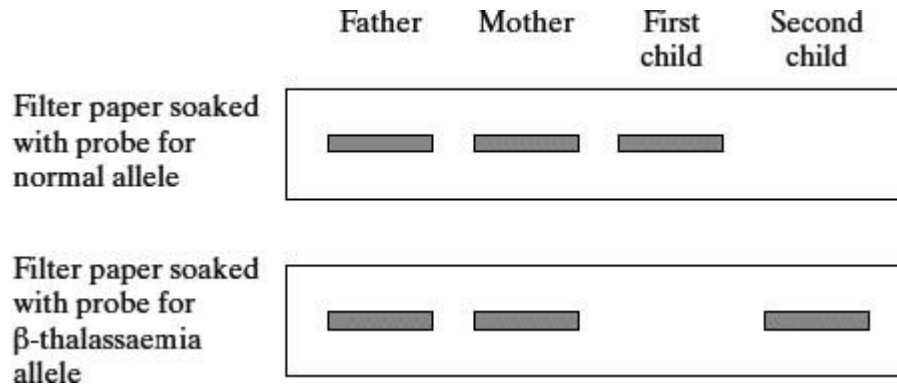
(Total 6 marks)

Q12.

β -thalassaemia is a genetic condition in which abnormal haemoglobin is produced. In one form, the recessive allele for β -thalassaemia, **t**, differs from the normal allele, **T**, by a single base-pair. A radioactive DNA probe was used to investigate the genotypes of four members of one family. The flowchart summarises the technique involved.



The diagram below shows the appearance of the two pieces of filter paper which resulted from the investigation.



- (a) What is the probability that the next child that this couple have is a girl who has β -thalassaemia? Explain your answer.

(3)

- (b) (i) The fragment of DNA containing the normal allele and the fragment with the β -thalassaemia allele moved the same distance on the gel. Explain why.

(2)

- (ii) The allele for β -thalassaemia differs from the normal allele by only one base-pair. Explain why the probe used to identify these alleles consists of a piece of DNA twenty bases in length and not just one base.

(2)

(Total 7 marks)

Q13.

Read the following passage.

Shark-fin soup is an expensive delicacy. To provide the basic ingredient, fishermen catch the sharks, hack the fins off and throw the dead bodies back into the ocean. But sharks are slow to mature and produce only a few offspring at a time, so they are vulnerable to overfishing. Monitoring the shark-fin trade is difficult, as once a fin has been cut off, it can be extremely difficult to work out precisely from which species it was taken.

The DNA from different species of sharks shows some differences in base sequence. This has enabled a new genetic fingerprinting technique to be developed. This technique would allow conservationists and fisheries managers to assess which of the 400 shark species are most threatened by the trade in shark fins.

An identification process has been developed using a range of “primers”. These are short pieces of single-stranded DNA that are complementary to a particular sequence of DNA. Each primer is specific to the DNA of one shark species.

The primers are added to DNA taken from a shark’s fin and the polymerase chain reaction is carried out. Only two primers, one at each end of a certain piece of DNA, will bind. The piece of DNA between the primers is replicated by the polymerase chain reaction. The primers that bind are specific to a particular species of shark and the length of the DNA fragment replicated differs for each species. When this DNA is run in an electrophoresis gel it produces a single band, enabling the researchers to identify which species of shark is involved.

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

- (a) (i) Explain why the DNA for each species of shark shows differences in base sequence (line 6).

(2)

- (ii) Each primer is specific to the DNA of one shark species (line 12).

Explain why a particular primer will only bind to the DNA of one species.

(2)

- (iii) The length of the replicated DNA fragment is different for each species.

Explain why this is important in identifying the shark species involved.

(3)

- Explain the reason for the difference in the number of 'rungs' produced.

(2)

- (6)

(Total 15 marks)

Soon a single drop of blood might be enough to reveal, at a very early stage, if a patient has

5 cancer. It could also tell us what type of cancer it is and whether it is treatable. Fragments of DNA from body cells are present in blood plasma. Some of these fragments may be from cancer cells. The fragments can be detected by a new test in which a test strip containing nucleic acid binds to sections of altered DNA.

Other cancer-detecting techniques involve removing a tissue sample from a patient. The tissue sample is used to obtain mRNA. By examining the mRNA, scientists can discover whether cancer is present.

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

(a) Describe how altered DNA may lead to cancer.

(6)

(b) Explain why fragments of DNA from cancer cells may be present in blood plasma (lines 3-4).

(2)

(c) Explain why the nucleic acid on the test strip will only bind to altered DNA (lines 4-5).

(2)

- (d) This test strip will allow cancers to be detected at a very early stage. Explain why cancer is more likely to be treated successfully if the disease is detected at a very early stage.

(2)

- (e) Explain how examining mRNA (line 7) enables scientists to discover whether cancer is present.

(3)

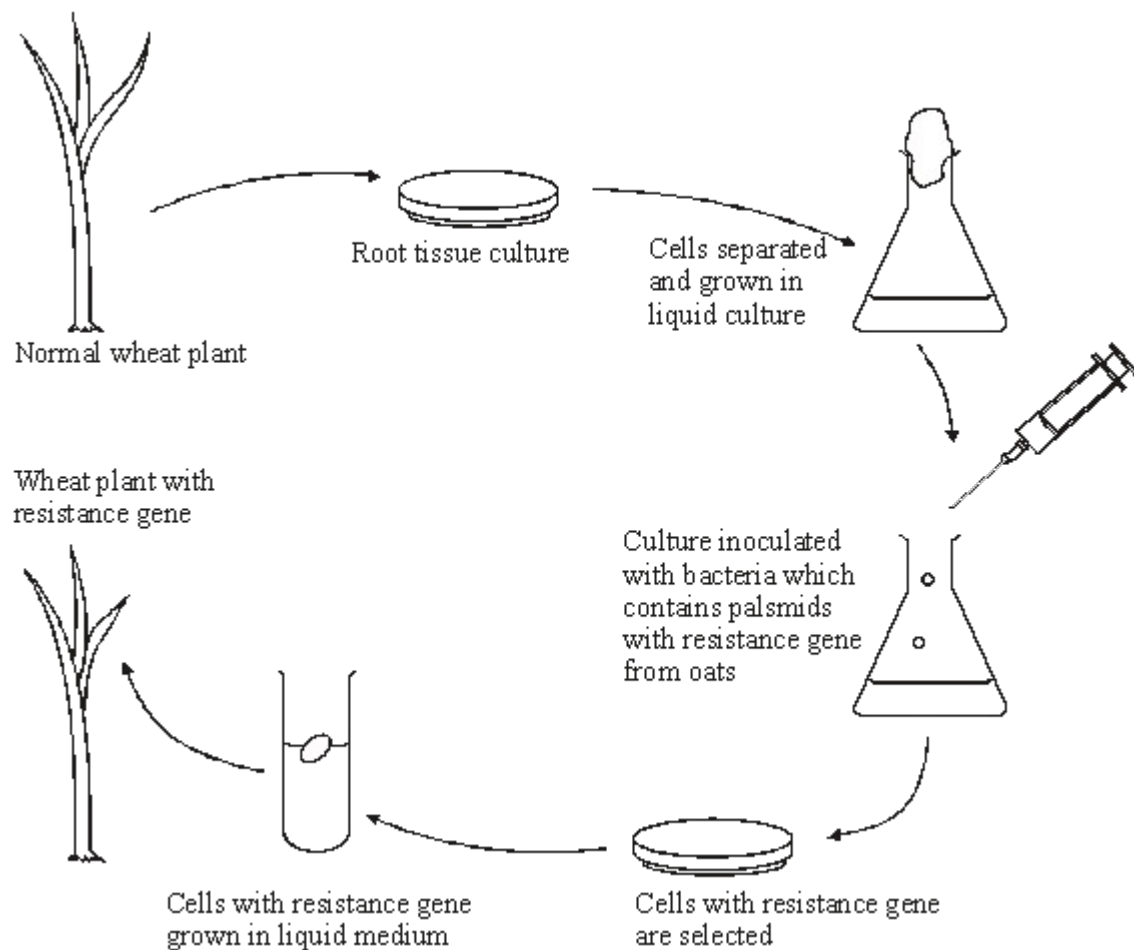
(Total 15 marks)

Q15.

'Take-all' is a disease of wheat caused by a fungus. It can cause serious damage to the crop.

There is no gene for resistance to this fungus in wheat. There is, however, a gene for resistance to this fungus present in oats.

The diagram shows how this gene might be transferred to wheat.



- (a) (i) The wheat plant with the resistance gene contains recombinant DNA. What is *recombinant DNA*?

(1)

- (ii) The plasmids act as vectors for the resistance gene. What is a *vector*?

(1)

- (iii) Suggest how cells with the resistance gene might be selected.

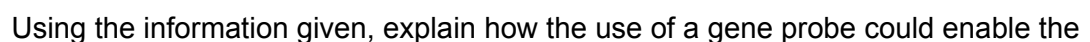
(2)

- (b) A laboratory has oat plants containing the resistance gene and a supply of plasmids.

This image shows a blank sheet of white paper with horizontal ruling lines. The lines are evenly spaced and extend across the width of the page. There are no margins, text, or other markings on the paper.

(Total 10 marks)

(a) Cystic fibrosis can be caused by any one of several mutant alleles of the cystic fibrosis gene. The most common of these mutant alleles accounts for about 70% of cases of cystic fibrosis. The use of gene probes can identify individuals carrying this allele. Gene probes are single strands of DNA which are radioactively labelled. They have a base sequence that is complementary to a mutant allele. The main stages in using a gene probe are shown in the diagram.



presence of a mutant allele of the cystic fibrosis gene to be detected.

(4)

- (b) Sheep have been genetically engineered to produce alpha-1-antitrypsin which is used to treat cystic fibrosis. Use your knowledge of this process to explain **one** argument for and **one** against using sheep in this way.

For

Against

(2)

(Total 6 marks)

Mark schemes

Q1.

- (a) 1. Correct answer of 19.4 / 19.41%
OR
19.47 / 19.5% = **2 marks**;
2. Incorrect answer but shows increase of
1,048,320 **OR** 1,051,200 = one mark;
Accept: 19.46% for one mark. 2
- (b) 1. Less / no acetylcholine broken down;
2. Acetylcholine attaches to receptors;
3. (More) Na⁺ enter to reach threshold / for depolarisation / action potential / impulse;
1. *Accept: more acetylcholine present / remains.*
1 and 2. *Accept: remains attached for longer = 2 marks.*
3. *Must be sodium ions.* 3
- (c) 1. Isolated **so** inbreeding / low genetic diversity / small gene pool;
2. Allele inherited (through generations) from (common) ancestor;
1. *Ignore: Founder effect.*
1. *Accept: no interbreeding with other populations.*
1. *Reject: interbreeding within the population.* 2
- (d) 1. AD / symptoms develops late / at 49;
2. Have already reproduced;
Note: 'It' is not equivalent to AD / symptom as the question stem relates to the mutation. 2
- (e) 1. Epigenetics / environment / named factor e.g. stress, alcohol, toxins, diet, exercise, smoking;
2. methylation (of genes)
OR
acetylation (of histones);
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.* 2
- (f) 1. One person was homozygous dominant / has two dominant alleles = **2 marks**;
2. For one mark has two alleles / chromosomes;
1. *Accept; homozygous dominant genotype e.g. 'one person has AA' for 2 marks.*
2. *Accept: is diploid or has two copies of the gene.* 2
- (g) 1. (GCA / triplet) is common / found in other places;

2. Would not know if it was the mutation / allele / gene

OR

Produces 'false positives'

1. *Accept: Probe will bind elsewhere.*

2

[15]

Q2.

- (a) (i) Does not code for amino acid/tRNA/rRNA;
Accept 'does not code for production of protein/polypeptide'
Reject 'that produces/makes amino acid'

1

- (ii) Deletion mutation;
Accept 'deletion'
Ignore references to splicing

1

- (b) (The) polymerase chain reaction;
Accept PCR

1

- (c) 1. Probes are single stranded / have a specific base sequence;
 2. Complementary base sequence on (specific) spacer

OR

3. Complementary/specific to (particular) spacer;
 4. (In white squares probe) binds (to single-stranded spacer) and glows/produces light/fluoresce;
2. Need idea of complementary to spacer
3. Accept converse for dark squares

3

- (d) 1. To see if strain is resistant to any antibiotics;
 2. So can prescribe effective/right antibiotic;

OR

3. To see whether (any) vaccine works against this strain/
 see which vaccine to use/ to produce specific vaccine;
 4. (So) can vaccinate potential contacts/to stop spread;

OR

5. Can test other people to see if they have the same strain/ to trace where people caught TB;
 6. Allowing control of spread of disease/vaccinate/treat contacts (of people with same strain) before they get TB;

Do not allow mix and match of points from different alternative pairs

2 max

[8]

Q3.

- (a) 1. Cut (DNA) at same (base) sequence / (recognition) sequence;
Accept: cut DNA at same place
2. (So) get (fragments with gene) **R** / required gene.
Accept: 'allele' for 'gene' / same gene 2
- (b) 1. Each has / they have a specific base sequence;
2. That is complementary (to allele r or R).
Accept description of 'complementary' 2
- (c) 1. Fragments L from parent rr, because all longer fragments / 195 base pair fragments;
Ignore: references to fragments that move further / less, require identification of longer / shorter or 195 / 135
Accept: (homozygous) recessive
2. Fragments N from parent RR, because all shorter fragments / 135 base pair fragments;
1 and 2 Accept: A3 for 195 and A4 for 135
2. Accept: (homozygous) dominant
3. (M from) offspring heterozygous / Rr / have both 195 and 135 base pair fragments.
Accept: have both bands / strips
Reject: primer longer / shorter 3
- (d) 1. (Cells in mitosis) chromosomes visible;
2. (So) can see which chromosome DNA probe attached to. 2
- (e) (i) 1. For comparison with resistant flies / other (two) experiments / groups;
Ignore: compare results / data / no other factors
2. To see death rate (in non-resistant) / to see effect of insecticide in non-resistant / normal flies.
Accept: 'pesticide' as 'insecticide'
Accept to see that insecticide worked / to see effect of enzyme 2
- (ii) (PM must be involved because)
1. Few resistant flies die (without inhibitor);
 2. More inhibited flies die than resistant flies;
 3. (PM) inhibited flies die faster (than resistant flies);
- (Other factors must be involved because)
4. Some resistant flies die;
 5. But (with inhibitor) still have greater resistance / die slower than non-resistant flies.
- Accept: (with inhibitor) die slower than non-resistant flies*

4 max

Q4.

- (a) Reverse transcriptase; 1
- (b) 1. Probe (base sequence) complementary (to DNA of allele A / where A is (and) binds by forming base pairs / hydrogen bonds;
Accept gene A
2. So (only) this DNA labelled / has green dye / gives out (green) light;
Accept glows for green light 2
- (c) (i) 1. More probe binding / more cDNA / mRNA / more allele / gene A means more light;
2. DNA (with **A**) doubles each (PCR) cycle;
3. So light (approximately) doubles / curve steepens more and more (each cycle) / curve goes up exponentially / increases even faster; 3
- (ii) (**G** because)
1. (Heterozygous) only has half the amount of probe for **A** attaching / only half the amount of DNA / allele A (to bind to);
Accept only one A to bind to
2. (So,) only produced (about) half the light / glow / intensity (of **H**) (per cycle of PCR);
If reference to 'half' for point 1, allow 'less light' in 2. 2

[8]

Q5.

- (a) 1. Carriers are heterozygous / have one normal copy and one mutant copy of gene / have one recessive allele / don't have the condition;
2. Both have DNA that binds (about) half / 50% amount of probe (that non-carrier does);
3. Probe binds to dominant / healthy allele so only one copy of exon in their DNA / have one copy of gene without exon / base sequence for probe to bind to;
3. *Accept normal and gene*
3. *Accept have a deletion mutation* 3
- (b) 1. Introns not translated / not in mRNA / (exons) code for amino acids / introns do not code for amino acids;
1. *Accept not expressed*
1. *Accept polypeptide / protein for amino acids*

2. Mutations of these (exons) affect amino acid sequences (that produce) faulty protein / change tertiary structure of protein;
 2. *Accept deletion leads to frameshift*
 2. *In this context, accept affects protein made*
3. So important to know if parents' exons affected, rather than any other part of DNA / introns;
 Accept converse arguments involving - eg introns do not code for amino acids / proteins
 Reject references to making amino acids, once

3

- (c) 1. Restriction mapping / described;
2. DNA / base sequencing (of fragments) / description / name of method;

2

[8]

Q6.

- (a) (i) 1. Negative correlation;
 Accept: description for 'negative correlation'
 Neutral: 'correlation'
 Reject: positive correlation
2. Wide range;
3. Overlap;
4. (Graph suggests that) other factors may be involved (in age of onset);
 2 / 3 *Accept the use of figures from the graph*
 2 / 3 *Can refer to age of onset or number of CAG repeats*
 Ignore references to methodology
- (ii) 1. Age of onset can be high / symptoms appear later in life;
 Accept: 'gene' for 'allele'
2. (So) individuals have already had children / allele has been passed on;

3 max

OR

3. Individuals have passed on the allele / already had children;
4. Before symptoms occur;

2 max

- (b) (i) 1. Person **K**;
2. (As has) high(est) band / band that travelled a short(est) distance / (er) so has large(st) fragment / number of CAG repeats;
 Must correctly link distance moved and fragment size

2

- (ii) Run fragments of known length / CAG repeats (at the same time);
Accept: references to a DNA ladder / DNA markers
Do not accept DNA sequencing
- (iii) Homozygous / (CAG) fragments are the same length / size / mass;
Accept: small fragment has run off gel / travelled further

1

1

[9]

Q7.

- (a) 1. Closer the (amino acid) sequence the closer the relationship;
2. (Protein structure) related to (DNA) base / triplet sequence;
Amino acid sequence is related to (DNA) base / triplet sequence = two marks;
- (b) 1. Reference to base triplets / triplet code / more bases than amino acids / longer base sequence than amino acid sequence;
Different (base) triplets code for same amino acids = 2 marks;
Degeneracy of triplet code = 2 marks
2. Introns / non-coding DNA / degeneracy of code / more than one code for each amino acid;
Ignore reference to codon.

2

2

[4]

Q8.

21 – 25	Extended abstract Generalised beyond specific context	Response shows holistic approach to the question with a fully integrated answer which makes clear links between several different topics and the theme of the question. Biology is detailed and comprehensive A-level content, uses appropriate terminology, and is very well written and always clearly explained. No significant errors or irrelevant material. For top marks in the band, the answer shows evidence of reading beyond specification requirements.
16 – 20	Relational Integrated into a whole	Response links several topics to the main theme of the question, to form a series of interrelated points which are clearly explained. Biology is fundamentally correct A-level content and contains some points which are detailed, though there may be some which are less well developed, with

		appropriate use of terminology. Perhaps one significant error and, or, one irrelevant topic which detracts from the overall quality of the answer.
11 – 15	Multistructural Several aspects covered but they are unrelated	Response mostly deals with suitable topics but they are not interrelated and links are not made to the theme of the question. Biology is usually correct A-level content, though it lacks detail. It is usually clearly explained and generally uses appropriate terminology. Some significant errors and, or, more than one irrelevant topic.
6 – 10	Unistructural Only one or few aspects covered	Response predominantly deals with only one or two topics that relate to the question. Biology presented shows some superficial A-level content that may be poorly explained, lacking in detail, or show limited use of appropriate terminology. May contain a number of significant errors and, or, irrelevant topics.
1 – 5	Unfocused	Response only indirectly addresses the theme of the question and merely presents a series of biological facts which are usually descriptive in nature or poorly explained and at times may be factually incorrect. Content and terminology is generally below A-level. May contain a large number of errors and, or, irrelevant topics.
0		Nothing of relevance or no response.

Commentary on terms and statements in the levels mark scheme

The levels mark scheme for the essay contains a number of words and statements that are open to different interpretations. This commentary defines the meanings of these words and statements in the context of marking the essay. Many words and statements are used in the descriptions of more than one level of response. The definitions of these remain the same throughout.

Levels mark scheme word/statement	Definition
Holistic	Synoptic, drawing from different topics (usually sections of the specification)
A fully integrated answer which makes clear links between several different topics and the theme of the question	All topics relate to the title and theme of the essay; for example, explaining the biological importance of a process.

	When considering, for example, the importance of a process, the explanation must be at A-level standard. ‘Several’ here is defined as at least four topic areas from the specification covered. This means some sentences, not just a word or two. It does not mean using many examples from one topic area.
Biology is detailed and comprehensive A-level content, uses appropriate terminology, and is very well written and always clearly explained.	Detailed and comprehensive A-level content is the specification content. Terminology is that used in the specification. Well written and clearly explained refers mainly to biological content and use of terminology. Prose, handwriting and spelling are secondary considerations. Phonetic spelling is accepted, unless examiners are instructed not to do so for particular words; for example, glucagon, glucose and glycogen.
No significant errors or irrelevant material.	A significant error is one which significantly detracts from the biological accuracy or correctness of a described example. This will usually involve more than one word. Irrelevant material is several lines (or more) that clearly fails to address the title, or the theme of the title.
For top marks in the band, the answer shows evidence of reading beyond specification requirements.	An example that is relevant to the title and is not required in the specification content. The example must be used at A-level standard.
Response mostly deals with suitable topics but they are not interrelated and links are not made to the theme of the question.	Not addressing the biological theme of the essay (e.g. importance) at <u>A-level standard</u> .

Please note that to obtain full credit, students must use information to show **the importance of Using DNA in science and technology**.

Topics

DNA and classification	
2.2	Structure of DNA

2.3	Differences in DNA lead to genetic diversity
2.9	Comparison of DNA base sequences
Genetic engineering and making useful substances	
2.5	Plasmids
5.8	The use of recombinant DNA to produce transformed organisms that benefit humans
Other uses of DNA	
2.5	Cell cycle and treatment of cancer
5.8	Gene therapy; Medical diagnosis and the treatment of human disease; The use of DNA probes to screen patients for clinically important genes.

In order to fully address the question and reach the highest mark bands students must also include at least four topics in their answer, to demonstrate a synoptic approach to the essay.

Students may be able to show the relevance of other topics from the specification.

Note, other topics from beyond the specification can be used, providing they relate to the title and contain factually correct material of at least an A-level standard. Credit should not be given for topics beyond the specification which are below A-level standard.

[25]

Q9.

- (a) (i) To cut the DNA;
Reject breakdown, cutting out 1
- (ii) To separate the (pieces of) DNA; 1
- (b) Complimentary base sequence / complementary DNA; binds to both (haplotypes);
Label would show up in both;
Idea of complementarity required 2
- (c) (i) Y chromosome inherited / comes from male parents / only found in males; 1
- (ii) Mitochondria in egg / female gamete / no mitochondria come from sperm / male gamete; 1
- (d) (i) Allows comparison;

Different (sized) areas covered;	2	
(ii) Wolves do not eat all of prey animal / do not eat (large) bones / skin; Inedible parts make up different proportions / wolf eats different proportions;	2	
(e) Limited by food / prey; as prey increases so do wolf numbers / positive correlation; Large range so other factors involved;	2	[12]

Q10.

(a) Restriction (enzyme / endonuclease);	1	
(b) Move towards anode / move because charged; Different rates of movement related to charge / size;	2	
(c) (i) Piece of DNA; Single stranded; Complementary to / binds to known base sequence / gene;	max 2	
(ii) DNA invisible on gel / membrane; Allows detection;	2	[7]

Q11.

(a) Endonuclease / restriction enzyme;	1	
(b) DNA made of base pairs; Each base pair is same length / occupies same distance along backbone;	2	
(c) (i) Second blank box from left labelled 6;	1	
(ii) Distance moved depends on length / number of base pairs / second longest fragment / second shortest distance identified;	1	
(d) 5;	1	[6]

Q12.

- (a) Mother and father both heterozygotes / Tt / carriers;
Probability of thalassaemia 1/4 and female 1/2;
Probability of both 1/8; 3
- (b) (i) Cut at same base sequence as same enzyme used;
Fragments are same length / size / have same charge; 2
- (ii) Single base occurs many times;
Sequence of 20 unlikely to occur elsewhere;
Allow one mark for establishing the principle where neither marking point clearly made. 2

[7]

Q13.

- (a) (i) Different genes / characteristics / features;
Reference to mutations;
Or
Base sequence determines protein;
Different species have different protein sequences; max 2
- (ii) Primer has different DNA sequence;
DNA specific / complementary base-pairing; 2
- (iii) Electrophoresis separates DNA;
(So they can be) identified by position on gel;
Smaller / shortest fragments travel furthest / quicker / or
reverse argument; 3
- (b) (*conventional*) Many lengths / all DNA / (*new*) one length;
Each rung is DNA of one / specific length; 2
- (c) 1 Heat DNA;
2 Breaks hydrogen bonds / separates strands;
3 Add primers;
4 Add nucleotides;
5 Cool;
6 (to allow) binding of nucleotides / primers;
7 DNA polymerase;
8 Role of (DNA) polymerase;
9 Repeat cycle many times; max 6

[15]

Q14.

- (a) 1 (DNA altered by) mutation;
2 (mutation) changes base sequence;
3 of gene controlling cell growth / oncogene / that monitors cell division;
4 of tumour suppressor gene;
5 change protein structure / non-functional protein / protein not formed;

- 6 (tumour suppressor genes) produce proteins that inhibit cell division;
- 7 mitosis;
- 8 uncontrolled / rapid / abnormal (cell division);
- 9 malignant tumour;

max 6

- (b) cancer cells die / break open;
releasing DNA;
- (c) normal DNA and changed DNA have different sequences;
DNA only binds to complementary sequence;
- (d) fewer abnormal / cancerous cells / smaller tumours;
less cell damage / less spread / fewer locations to treat;
- (e) mRNA base sequence has changed;
gene / DNA structure is different / has mutated;
cancer gene active / tumour suppressor gene inactive;

2

2

2

3

[15]

Q15.

- (a) (i) contains genes / nucleotides / sections of DNA / artificial
DNA from two species / 2 types of organisms;
- (ii) carries gene / DNA (into the other organism / gene carrier);
- (iii) expose cells to the fungus;
non-resistant ones die, resistant ones survive;
OR identify by adding marker gene / gene probe / (qualified)
marker probe; description of positive result
e.g. radioactivity / fluorescence / complementary base pairing;

1

1

2

- (b) EITHER 1 cut desired gene (from DNA) of oat plant;
2 using restriction endonuclease / restriction enzyme;
OR 1 use mRNA from oat which will code for resistance;
2 and use reverse transcriptase to form desired DNA;
OR 1 make artificial DNA with correct sequence of bases;
2 using DNA polymerase;
3 cut plasmid open;
4 with (same) restriction endonuclease / restriction enzyme;
5 ref. sticky ends / unpaired bases attached;
6 use (DNA) ligase to join / ref. ligation;
7 return plasmid to (bacterial) cells;
8 use of Ca^{2+} / calcium salts / electric shock;
(if ref. to 'insulin' allow 5 max.)

max 6

[10]

Q16.

- (a) probe will attach (to mutant allele);
attaches to one DNA strand;
as a result of complementary base pairing;
radioactivity detected on film / X-ray / by autoradiography
(if mutant allele present);

4

- (b) for
gene is only active in mammary cells / only affects milk / easy to
obtain product / product produced in large amounts / gene passed to
offspring;

1

against

long term effects not known / qualified reference to animal exploitation
e.g. use of embryos / effect of inserted gene on other sheep
tissues / genes;

1

[6]

Examiner reports

Q1.

- (a) Over a third of students obtained both marks for this question, for answers of 19.41 / 19.4% or 19.47 / 19.5%, depending on whether the student used 7 x 52 (weeks) or 365 as the number of days in a year. Almost a third of students gained one mark for correctly calculating the increase in AD cases per year as being 1 048 320 or 1 051 200, depending on the number of days used. Incorrect rounding to give 19.46% was quite common, to gain one mark.
- (b) The majority of students gained at least one mark for stating that less acetylcholine would be broken down, or that more acetylcholine would be present. Almost half of these students obtained a second mark for stating that the acetylcholine binds to receptors. However, only 10% of students obtained maximum marks by describing how an impulse would be produced in the postsynaptic neurone. Many students did appreciate that sodium ion channels would open, but then failed to mention that sodium ions would then enter to cause depolarisation.
- (c) Over 50% of students scored zero for this question, usually due to answers lacking complete explanations for the valid suggestions they outlined. Less than 2% of students gained both marks. The most frequently credited response was that isolation had resulted in a small gene pool or low genetic diversity. Poor use of terminology also prevented many students gaining both marks. Invariably, students referred to the gene or mutation, rather than the allele being inherited from a (common) ancestor. There was also considerable confusion in the use of the terms inbreeding and interbreeding. There were also many responses which referred to genetic bottlenecks, the Founder effect, and an increase in the rate of mutation in isolated areas. A significant number of students suggested that the late onset of AD enabled individuals still to reproduce and pass on the mutation. This would explain why the frequency of the mutation had not been reduced (part d), rather than why there is a high frequency of this mutation to begin with.
- (d) Almost a third of students obtained both marks, clearly expressing the idea that, due to the symptoms of AD developing late on, affected individuals would have already reproduced. Over 50% of students scored zero, often providing responses that suggested that the mutation was not harmful, or indeed that it was beneficial. An improvement in health care was also provided as an explanation for the frequency of the mutation not being reduced. Students obtaining one mark often did not refer to the late onset of AD, but did understand that individuals with the mutation could still reproduce and pass on the allele.
- (e) Over 50% of students scored zero on this question. Many of these responses suggested that differences in the 'level' of acetylcholine, or exposure to mutagenic agents, caused the variation in the age at which the mutation is expressed. Over a third of students obtained one mark, usually by naming an environmental factor such as diet, smoking or stress. Answers specifically referring to epigenetics for at least one mark were infrequent. These responses often gained a second mark, 10% of students, for mentioning methylation or acetylation. Students who described these processes generally provided correct details.
- (f) Almost three out of ten students obtained one mark for explaining the sample size of 204 in terms of two copies of chromosome 14 or two copies of an allele. A common error was to refer to two chromatids. Explaining why there were only 74 potential AD cases when 75 mutations had been detected proved very challenging, with only 2% of students gaining this second mark. Many students suggested that the allele

causing AD is recessive, despite line 18 of the comprehension passage stating that it is dominant. A common misconception was that one individual was heterozygous for the condition. Other incorrect responses focused on AD not having yet developed, or attempted to explain the data in relation to the degeneracy of the genetic code.

- (g) Over 10% of students did not attempt this question, and over 5% omitted the parts (e) and (f). It seems likely that some students had difficulty completing the paper, but it was also evident that these last three questions were demanding. Only 25% of students obtained a mark on this question. Almost all of these students gained one mark for realising that the GCA triplet would occur in a number of different places. Half of these students then explained that you could not then determine if the mutation was present or not. As in part (f), a number of incorrect ideas were linked to the degenerate nature of the genetic code. The misconception that probes were being used to sequence the whole genome arose, so lots of different primers would be needed and it would be very time consuming and costly. The probe was sometimes thought only to be able to identify the mutation if the gene had been expressed to cause the disease, or that the probe would not bind because the mutation had not occurred yet. A surprising number of students said the sequence of the mutation was not known, so therefore a probe could not be made, or that the mutation was different in different people.

Q2.

- (a)
 - (i) 72% correctly wrote about not coding for amino acids. Some failed to score because they wrote about 'not producing/making amino acids'.
 - (ii) Most students ignored the fact that the DR region is in DNA and wrote about splicing/removing the non-coding sequence from mRNA. The other 22% wrote, correctly, about deletion mutations.
- (c) This question discriminated well across the range of marks. 50% obtained two marks, usually for reference to a probe binding to a complementary base sequence on spacer DNA and then linking this to production of light. Only 13% also stated that each probe has a specific base sequence. 14% failed to score. Some referred to complementary base pairing, rather than sequences.
- (d) Surprisingly, 40% scored zero. There were many very vague answers about picking the right treatment and not encouraging resistance. 20% obtained both marks, usually for the idea of identifying any antibiotics to which the strain was resistant, so an appropriate antibiotic can be selected. The other mark points relating to vaccines and tracing sources of infection were also seen.

Q3.

- (a) Over half of students managed to communicate the idea that the same restriction enzymes cut at the same place / recognition sequence on DNA. Only just over 10 percent considered the context of the question and went on to say that this would give fragments containing the same gene (R).
- (b) About half of students obtained both marks in this part and over 40 percent obtained 1. Most managed to convey the idea that there was binding between complementary base sequences on a primer and DNA from an allele. The better answers conveyed the idea of a specific base sequence for each primer.
- (c) Some very good and clear answers were seen to this part and a third of students obtained 3 marks. These students identified the genotypes of L, M and N and

explained how they identified them on the basis of the sizes of primer attached to each and how far the bands moved. Quite a large number went on at length about the offspring as represented by M but failed to identify L and N. A few thought that there were two types of offspring because there were two bands for M; they clearly did not understand the simple genetics of the cross.

- (d) This proved far more challenging than expected. The examiners were looking for the idea that chromosomes would be visible (as separate structures) and thus the scientists could see to which chromosome the probe was attached. Fewer than 10 percent got both marks.
- (e)
 - (i) There were quite a large number of attempts at generic 'How Science Works' answers to this part. 50 percent of students obtained 1 mark, usually for suggesting that the control was to see the effect of the insecticide or that it was for comparison with resistant flies. However, only around 10 percent gave both of these.
 - (ii) Most students focused on one aspect of the results and scored 2 marks. They frequently spotted that if the enzyme was the only factor in resistance, then the results for the controls and the resistant flies with the inhibitor would be the same. Others focused on evidence for the statement by comparing the resistant flies with resistant flies with the inhibitor added.

Q4.

- (a) About 80% of students identified the enzyme in this part as reverse transcriptase. The commonest wrong answer appeared to be restriction endonuclease.
- (b) Most students obtained one mark for stating that the DNA probe has a base sequence complementary to the DNA of allele A. A third obtained a second mark by going on to state that this allowed it to bind to the target DNA by base pairing, or that this meant only target DNA gives off green light.
- (c)
 - (i) About half of students obtained one mark in this part for noting that the more probe binding to allele A, the more green light there would be. Nearly a quarter obtained a second mark, usually for also noting that the light curve goes up exponentially (or described). Only a few, 14%, obtained all three marks. These students explained that this was because the amount of DNA doubles (approximately) with each PCR cycle.
 - (ii) Answers to this part often made references to G being heterozygous because this person had *fewer* A alleles and thus *less* light was produced. The examiners were looking for more precise statements relating to half the amount of A with probe attached and half (approximately) the light produced (at any given time). A third of students obtained both marks.

Q5.

- (a) To answer this question, students had to appreciate that a person has two copies of each gene but in a carrier of a condition, the alleles are different. Many students appeared not to have grasped this concept. Just over one-third of students obtained one mark. The commonest mark awarded was for noting that the parents' DNA bound (about) half the amount of DNA probe that bound to the DNA of the person not carrying the mutation. Many went on to say that the parents were heterozygous (or described this). Relatively few stated that the DNA probe binds to the non-mutant form / allele of the gene. There were a lot of vague responses about less probe binding and fewer genes, or less DNA binding to probe in the parents.

- (b) Many students obtained one mark for noting that introns are spliced out of pre-mRNA to form mRNA. Others obtained a mark for stating that only exons code for amino acids. Some obtained marks for noting that only mutations of exons would affect amino acid sequences, or affect the protein produced. In general, few students gave the whole story. Some students wrote about exons producing amino acids and this was not given credit. Others strayed into (often long and inaccurate) accounts of introns and genetic fingerprinting.
- (c) This question produced a lot of partial accounts of how to sequence DNA.

Q6.

- (a)
 - (i) Although the negative correlation was usually indicated, only the better students appreciated the wide range in the age of onset or the overlap in values. Very few students were aware that the wide range in age of onset for the same number of CAG repeats suggests that other factors may also be involved.
 - (ii) A minority of weaker students gave answers that were out of context. Some thought that the allele for Huntington's disease is recessive and would therefore be passed on to offspring without a person knowing. Others thought that people with Huntington's disease would survive well into adulthood and then reproduce.
- (b)
 - (i) Most students correctly identified Person **K** as testing positive for Huntington's disease. They went on to explain that this person has the fragment that moved the shortest distance and linked this to a greater number of CAG repeats. A minority of students failed to link correctly the distance moved with the length of the fragment.
 - (ii) Students who failed to gain credit often referred to using DNA sequencing or probes to highlight sequences and 'restriction mapping'.
 - (iii) Misconceptions seen in responses by weaker students included partial digestion of DNA, Person **L** only having one allele of this gene and the probe not being able to attach to the other fragment.

Q7.

- (a) Over 40% of students failed to score on this question. Many of these students suggested that proteins consist of bases and the confusion between bases and amino acids pervaded their responses. Although a number of students did correctly refer to the sequence of amino acids, only better students linked the similarity of the amino acid sequence with a close evolutionary relationship between different species.
- (b) This question also proved challenging with less than 50% of students gaining any marks. However, a significant number of students did gain one mark for reference to the triplet code and students appreciating the degeneracy of this code were able to gain both marks. Some students gained credit by referring to introns or non-coding DNA.

Q8.

Using DNA in science and technology

The very best essays from candidates who selected this option were outstanding. They

reviewed, often in great detail, the relevant aspects of the specification although not always incorporating the role of DNA in the classification of organisms. Considering that much of the content of this essay could be drawn from this unit, it was surprising how poor many answers were. Understanding of techniques was often extremely limited, particularly *in vivo* gene cloning and the use of markers. Many essays presented no more than a broad overview either emphasising ethical issues at the expense of biological detail or failing to distinguish established practice from wishful thinking.

Q9.

- (a) Most candidates had some understanding of the function of restriction endonuclease but were not always sure of its role in the investigation described. Thus, there were numerous references to the enzyme “cutting out” particular sections of DNA, these pieces ranging from haplotypes, to genes and even chromosomes. Most candidates correctly suggested that electrophoresis would be involved in separating the DNA fragments, although some were clearly of the opinion that it was the chains of DNA that were separated.
- (b) Candidates were generally able to describe the complementary base sequence present on the probe but seldom progressed to explain how it could be used to show that the haplotypes concerned were the same.
- (c) The majority of candidates linked the Y-chromosome to male inheritance in part (i) although a significant number suggested that the Y-chromosome was inherited from the female. Part (ii) was targeted at stronger candidates, but very few could suggest that mitochondria could only be passed to the offspring in the cytoplasm of the egg.
- (d) The responses to part (i) suggested that while many candidates were aware that giving the units per unit area enabled comparison, they were uncertain as to what was being compared. The most frequent suggestion was that it allowed wolves to be compared with prey numbers. Others wrote about the territorial behaviour of wolves or suggested that the mobility of the animals made counting over a larger area too difficult. In part (ii), better candidates appreciated that wolves ate only part of their prey and that the amount eaten differed with different species of prey.
- (e) Although the positive correlation between prey index and wolf numbers was usually recognised, few progressed to state that this suggested that food must be limiting population size. Unfortunately, the few who pointed out that other factors might possibly be involved rarely linked this conclusion to the spread of data on the graph.

Q10.

- (a) Most identified the enzymes as restriction enzymes though the variety of spellings used meant that credit could not always be awarded.
- (b) This question showed the the idea of larger fragments moving slower or not as far was well understood although there was much incorrect biology relating to charge.
- (c) In (i) the idea of complementarity was understood and communicated well by many candidates although the fact the DNA probe is single stranded was not seen in many responses. In (ii) the majority of candidates obtained one of the available marks for the idea of detecting the location. Surprisingly few candidates stated that without a radioactive probe the location would not be visible. Many candidates had knowledge of fluorescent markers and gave a response based on this knowledge rather than answering the question. A common misconception was that radioactivity shows up when X-rayed.

Q11.

Unit 2

- (a) Most had no trouble here in identifying endonuclease.
- (b) This was badly done since few mentioned or implied that DNA was made of base pairs and that these occupied the same distance along the backbone. Many made it obvious that they meant the distance from one chain to the other or went on to write about the distances in the electrophoresis gel.
- (c) Although the correct box was often located, the second from the right was almost as commonly given. Appropriate justification for the choice was rewarded in (ii) although many candidates suggested fragment 6 was the fifth rather than the second largest.
- (d) All figures were given here with 5 only rarely chosen, 6 being most common.

Unit 3

In part (a), restriction enzyme was correctly identified by almost all candidates. However, part (b) attracted very poor answers. Few commented that DNA is composed of base pairs, or that all base pairs have the same length. Many simply re-stated the question. In (c) (i), most candidates correctly identified the box, but a significant number identified the penultimate box. These candidates explained in (c)(ii) that the *longest* fragments moved furthest. Although they had the correct idea that DNA separates according to length, they had the relationship the wrong way round. In (d), there were few correct answers. Most gave 6.

Q12.

- (a) Figure 2 showed that the male involved in this cross possessed two alleles and this should have alerted candidates to the fact that the gene concerned was not sex-linked. Most of those who identified it as being autosomal, were able to explain that the probability of the next child having thalassaemia was one in four. Rather less success was enjoyed, however, when it came to combining probabilities, and the mathematics of multiplying fractions was evidently beyond the ability of a number of candidates.
- (b) Part (i) was generally answered well by those candidates who distinguished between the processes of electrophoresis, chromatography and centrifugation. The basis of separation in this case is the difference in charge and, in DNA, this translates into differences in length. The numerous references to such features as solubility suggested that the distinction between these three processes was not always secure. The difficulties encountered by many in part (ii) stemmed largely from confusion between the terms allele, base and probe. There was, as a result, much inaccurate biology and little opportunity for awarding credit.

Q13.

This question proved to be very demanding for many candidates, although better candidates were able to score all, or almost all, of the available marks. In (a) (i), many candidates recognised that different species of shark have different characteristics, but mostly failed to link these characteristics to proteins or to genes. In (a) (ii), some candidates did realise that each primer had a specific base sequence which would bind to the shark DNA by complementary base-pairing, but weaker candidates simply re-stated the information in the stem. In (a) (iii), many candidates limited their answers to stating that the DNA fragment had to be a different length in each species so it could be

identified. How this enabled identification was not stated. On the other hand, good candidates wrote succinctly about electrophoresis separating DNA by size, and explained how the different length fragments would move different distances up the gel. Part (b) was poorly done by most candidates. It was common to find descriptions of genetic fingerprinting here. A few very weak students confused the 'rungs' with base pairs in the DNA molecule. Part (c) was the most accessible part of the question. A large proportion of candidates clearly knew the polymerase chain reaction and gave excellent accounts, often gaining full marks. A minority of very weak candidates confused PCR and transcription.

Q14.

- (a) Many candidates gave a good account of the changes a mutation could produce and those with clear expression achieved full marks; many scored three or four marks. Uncontrolled cell division and malignant tumors were frequently referred to and some appreciated that genes which controlled cell division could have changed. References to benign tumours or cell mutations were irrelevant in the context of this question.
- (b) Very few candidates achieved marks here, mainly because they did not read the question. Whole cells in the blood were not required, but the understanding that cancer cells could burst or die and release their DNA was.
- (c) Few seemed to understand this and restated the question without reference to the changed base sequences to which the strip would bind.
- (d) This was generally well known. The main reason for failing to gain marks was a reference to an undefined 'it' which would be growing, dividing or spreading, causing undefined damage.
- (e) Here too some candidates who understood the problem found it hard to explain that changes in the mRNA would reflect mutations in the DNA and would show that a cancer gene was active.

Q15.

- (a) Only the most able could explain that two species were required here for recombinant DNA although many could describe a vector. The third part elicited some thoughtful responses along the lines of exposing the cells to the fungus. Less able candidates wrote about exposure to the disease, and most went into antibiotic resistance without applying it to the situation here.
- (b) Many candidates were able to gain maximum credit here in a very few lines of script. However, there were still the misconceptions that genes are cut out of the plasmid and that this is 'splicing'. Some failed to address the context, or discussed fermenters.

Q16.

Not surprisingly, this question produced a wide range of marks. There were some excellent answers but even the most able candidates had difficulty obtaining maximum marks in part (a).

The vast majority of candidates obtained one or two of the four marks available. The first mark gained was by indicating that the gene probe would attach to the mutant allele. Some of these candidates obtained a second mark by stating that autoradiography could be used to detect radioactivity. However, a number of candidates suggested that 'firing X-rays' at the sample would suffice. Another misconception was that the individual

radioactive nucleotides would line up with the mutant allele rather than a single radioactive strand. Very few candidates appreciated that the gene probe would attach to one DNA strand and even fewer candidates explained that this would occur as a result of complementary base pairing, despite the cues given in the stem of the question.

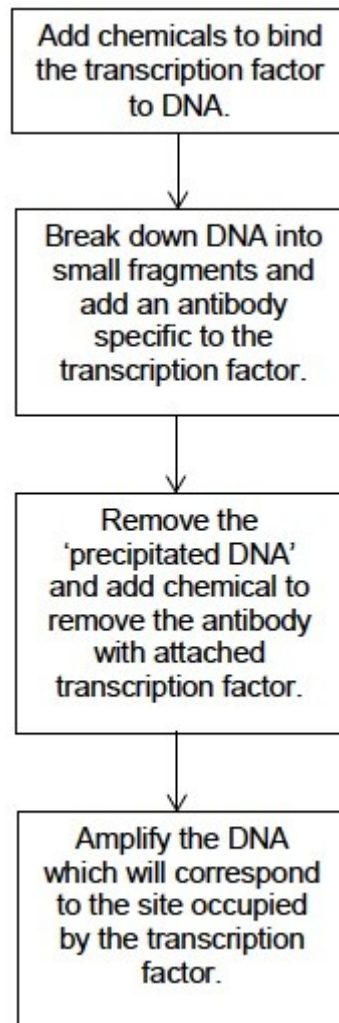
Only better candidates used their knowledge of the process involved to explain one argument for the use of genetically engineered sheep to produce alpha-1-antitrypsin. Generally these candidates referred to the product being in the milk or being produced in large quantities. Many candidates simply stated that you could treat cystic fibrosis, which was stated in the stem of the question. Fewer candidates gained a mark for explaining one argument against the use of these sheep. It was disappointing to see a large number of candidates simply referring to 'not playing God'. The most common correct response was to state that long-term effects are not known. Some candidates did refer to the low success rate of using sheep embryos in this process.

Q1.

- (a) What is meant by a genome?

(1)

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



- (b) Explain why the antibody binds to the transcription factor.

(2)

- (c) Use the chart to explain what 'precipitated DNA' consists of.

(1)

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.

- (d) Suggest how single-stranded cDNA could prevent transcription of the *P34* gene.

(1)

- (e) Describe the roles of **two** named types of enzymes used to insert DNA fragments into plasmids.

Type of enzyme _____

Role _____

Type of enzyme _____

Role _____

(2)

- (f) The soybean cells were screened for the presence of the P34 protein. This process involved the use of gel electrophoresis to separate proteins extracted from soybean cells.

Suggest **two** features of the structure of different proteins that enable them to be separated by gel electrophoresis.

1. _____

2. _____

(2)

(Total 9 marks)

Q2.

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

- (a) The scientists began by lysing (breaking open) cells and organelles using a detergent that dissolves lipids in water.

Suggest how the detergent releases CENP-W from cells.

(2)

- (b) Explain how ultracentrifugation separates CENP-W from other molecules.

Q3.

Some populations of flies are becoming resistant to insecticides intended to kill them.

Scientists developed a method for finding out whether a fly was carrying a recessive allele, **r**, that gives resistance to an insecticide. The dominant allele, **R**, of this gene does not give resistance.

The scientists:

- crossed flies with genotype **RR** with flies with genotype **rr**
- obtained DNA samples from the parents and offspring
- used the same restriction endonuclease enzymes on each sample, to obtain DNA fragments.

- (a) Explain why the scientists used the same restriction endonuclease enzymes on each DNA sample.

(2)

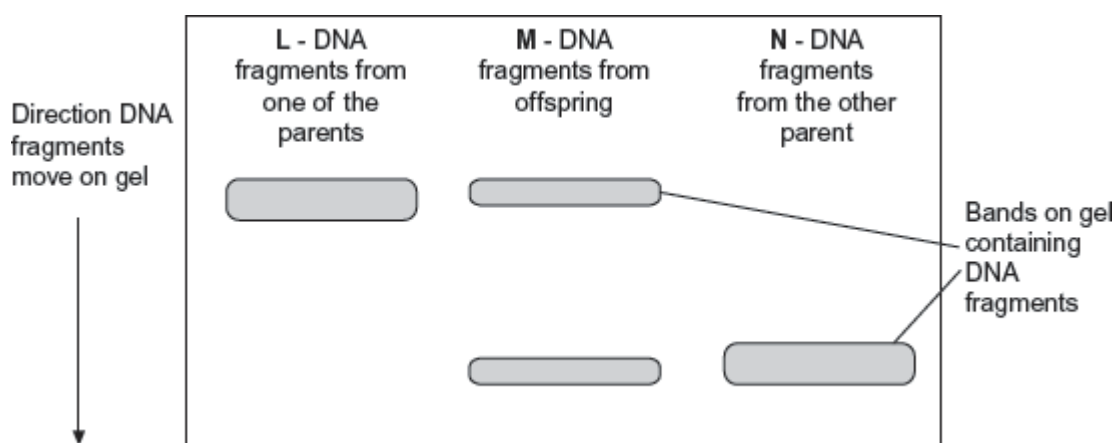
The scientists added two different primers to each sample of DNA fragments for the polymerase chain reaction (PCR).

- Primer A3 only binds to a 195 base-pair fragment from allele **r**.
- Primer A4 only binds to a 135 base-pair fragment from allele **R**.

The scientists separated the DNA fragments produced by the PCR on a gel where shorter fragments move further in a given time.

Their results are shown in **Figure 1**.

Figure 1



- (b) Explain why primer A3 and primer A4 only bind to specific DNA fragments.

(2)

- (c) Use all the information given to explain the results in **Figure 1**.

(3)

- (d) The scientists wanted to know on which chromosome the gene with alleles **R** and **r** was located. From the flies with genotype **RR**, they obtained cells that were in mitosis and added a labelled DNA probe specific for allele **R**. They then looked at the cells under an optical microscope.

Explain why they used cells that were in mitosis.

(2)

- (e) Another group of scientists thought that pesticide resistance in some flies was related to increased activity of an enzyme called P450 monooxygenase (PM). This enzyme breaks down insecticides.

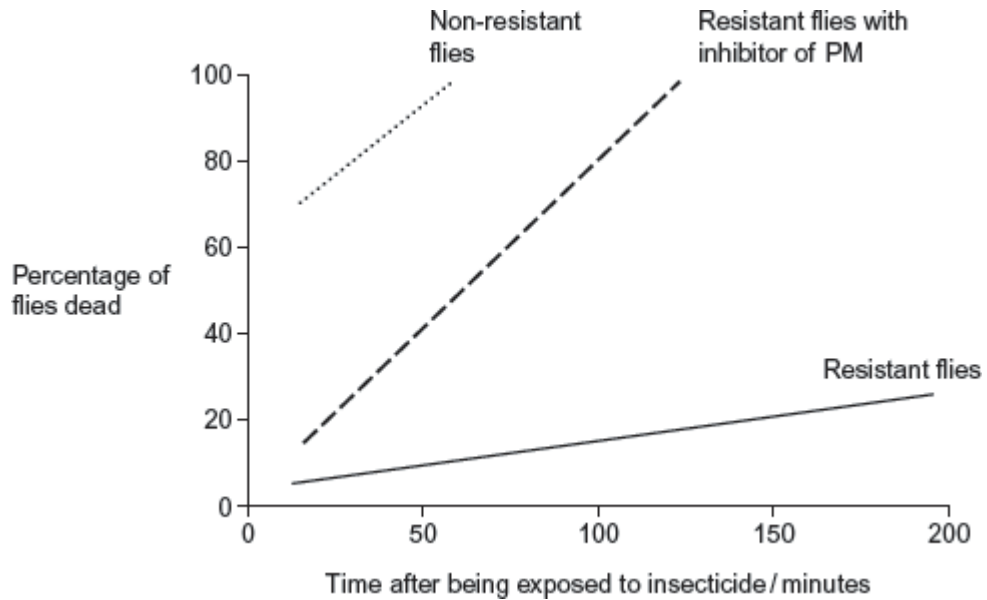
The scientists obtained large numbers of resistant and non-resistant flies. They then set up the following experiments.

- Non-resistant flies exposed to insecticide.
- Resistant flies exposed to insecticide.
- Resistant flies treated with an inhibitor of PM and then exposed to insecticide.

They then determined the percentage of flies that were dead at different times after being exposed to insecticide.

Figure 2 shows their results.

Figure 2



- (i) Explain why the scientists carried out the control experiment with the non-resistant flies.

(2)

- (ii) The scientists concluded that the resistance of the flies to the insecticide is partly due to increased activity of PM but other factors are also involved.

Explain how these data support this conclusion.

(4)

(Total 15 marks)

Q4.

Scientists wanted to measure how much mRNA was transcribed from allele **A** of a gene in a sample of cells. This gene exists in two forms, **A** and **a**.

The scientists isolated mRNA from the cells. They added an enzyme to mRNA to produce

cDNA.

- (a) Name the type of enzyme used to produce the cDNA.

(1)

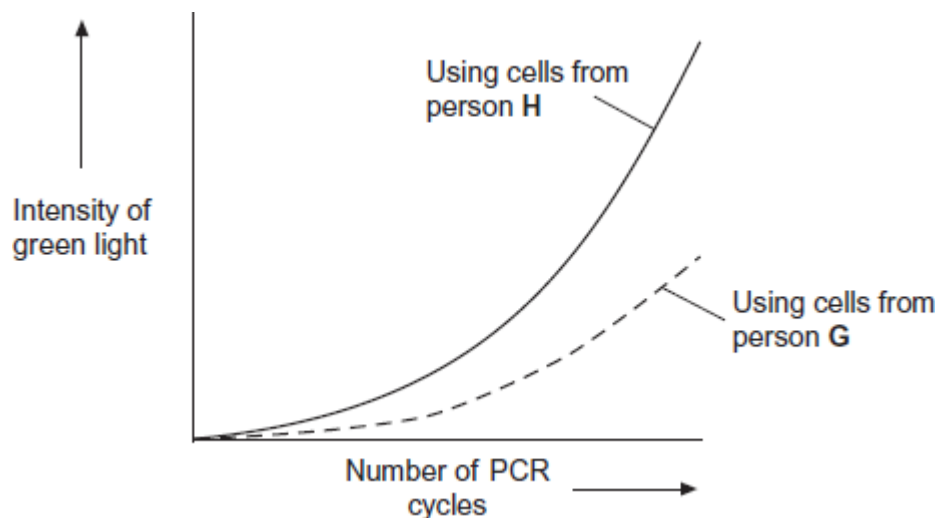
The scientists used the polymerase chain reaction (PCR) to produce copies of the cDNA. They added a DNA probe for allele **A** to the cDNA copies. This DNA probe had a dye attached to it. This dye glows with a green light **only** when the DNA probe is attached to its target cDNA.

- (b) Explain why this DNA probe will only detect allele **A**.

(2)

- (c) The scientists used this method with cells from two people, **H** and **G**. One person was homozygous, **AA**, and the other was heterozygous, **Aa**. The scientists used the PCR and the DNA probe specific for allele **A** on the cDNA from both people.

The figure shows the scientists' results.



- (i) Explain the curve for person **H**.

(3)

- (ii) Which person, **H** or **G**, was heterozygous, **Aa**? Explain your answer.

(2)

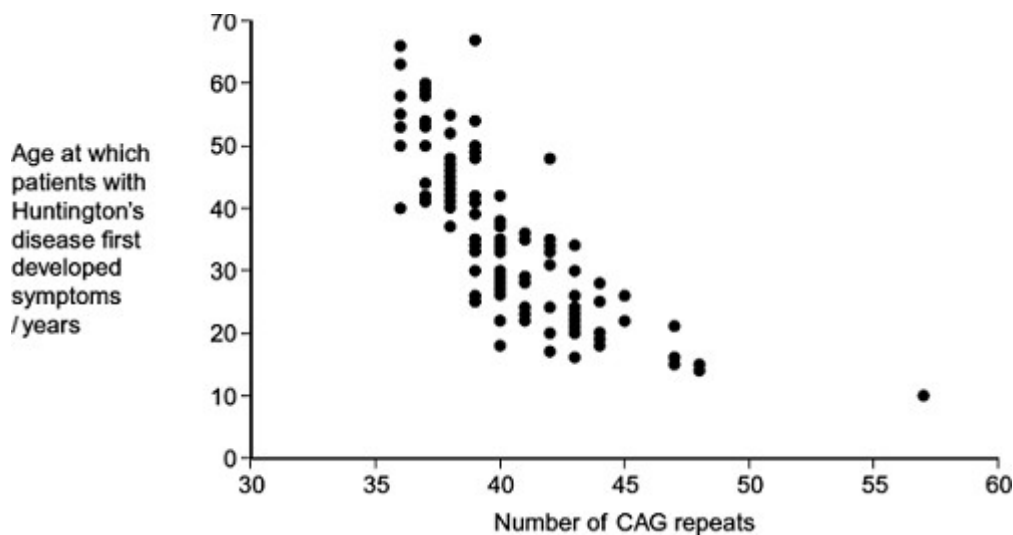
(Total 8 marks)

Q5.

Huntington's disease is a genetic condition that leads to a loss in brain function. The gene involved contains a section of DNA with many repeats of the base sequence CAG. The number of these repeats determines whether or not an allele of this gene will cause Huntington's disease.

- An allele with 40 or more CAG repeats will cause Huntington's disease.
- An allele with 36 – 39 CAG repeats may cause Huntington's disease.
- An allele with fewer than 36 CAG repeats will not cause Huntington's disease.

The graph shows the age at which a sample of patients with Huntington's disease first developed symptoms and the number of CAG repeats in the allele causing Huntington's disease in each patient.



- (a) (i) People can be tested to see whether they have an allele for this gene with more than 36 CAG repeats. Some doctors suggest that the results can be used to predict the age at which someone will develop Huntington's disease.

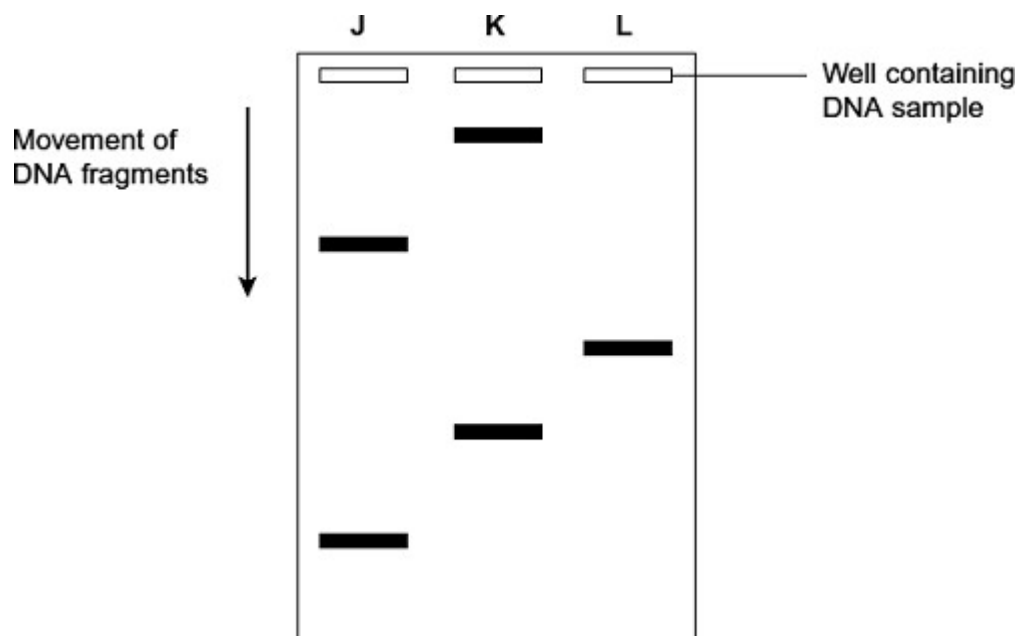
Use information in the graph to evaluate this suggestion.

(3)

- (ii) Huntington's disease is always fatal. Despite this, the allele is passed on in human populations. Use information in the graph to suggest why.

(2)

- (b) Scientists took DNA samples from three people, **J**, **K** and **L**. They used the polymerase chain reaction (PCR) to produce many copies of the piece of DNA containing the CAG repeats obtained from each person. They separated the DNA fragments by gel electrophoresis. A radioactively labelled probe was then used to detect the fragments. The diagram shows the appearance of part of the gel after an X-ray was taken. The bands show the DNA fragments that contain the CAG repeats.



- (i) Only one of these people tested positive for Huntington's disease. Which person was this? Explain your answer.

Person _____

Explanation _____

(2)

- (ii) The diagram only shows part of the gel. Suggest how the scientists found the number of CAG repeats in the bands shown on the gel.

(1)

- (iii) Two bands are usually seen for each person tested. Suggest why only one band was seen for Person L.

(1)

(Total 9 marks)

Q6.

Write an essay on using DNA in science and technology.

(Total 25 marks)

Q7.

There are wolves in many European countries. Scientists investigated the genetic diversity of these wolves. They collected samples of DNA from the mitochondria of wolves from different countries. For each sample they identified which haplotypes were present in the DNA. A haplotype is a particular sequence of bases on DNA. Mutations can produce new haplotypes.

Country	Number of wolves sampled	Number of different haplotypes in mitochondrial DNA
Spain	84	3
Portugal	19	2
Italy	101	1
France	7	1
Bulgaria	29	6
Sweden	93	1

The scientists wanted to find out whether one of the haplotypes in the Portuguese wolves was the same as one of those in the Spanish wolves. They used a restriction endonuclease, electrophoresis and a labelled DNA probe.

(a) For what purpose did they use

(i) the restriction endonuclease

(1)

(ii) electrophoresis?

(1)

(b) Explain why the labelled DNA probe could be used to find out whether the haplotypes were the same.

(2)

(c) The scientists analysed the DNA on the Y chromosome and the DNA in the mitochondria of the Swedish wolves. They concluded that the Swedish wolf population descended from one male wolf from Finland and one female wolf from Russia.

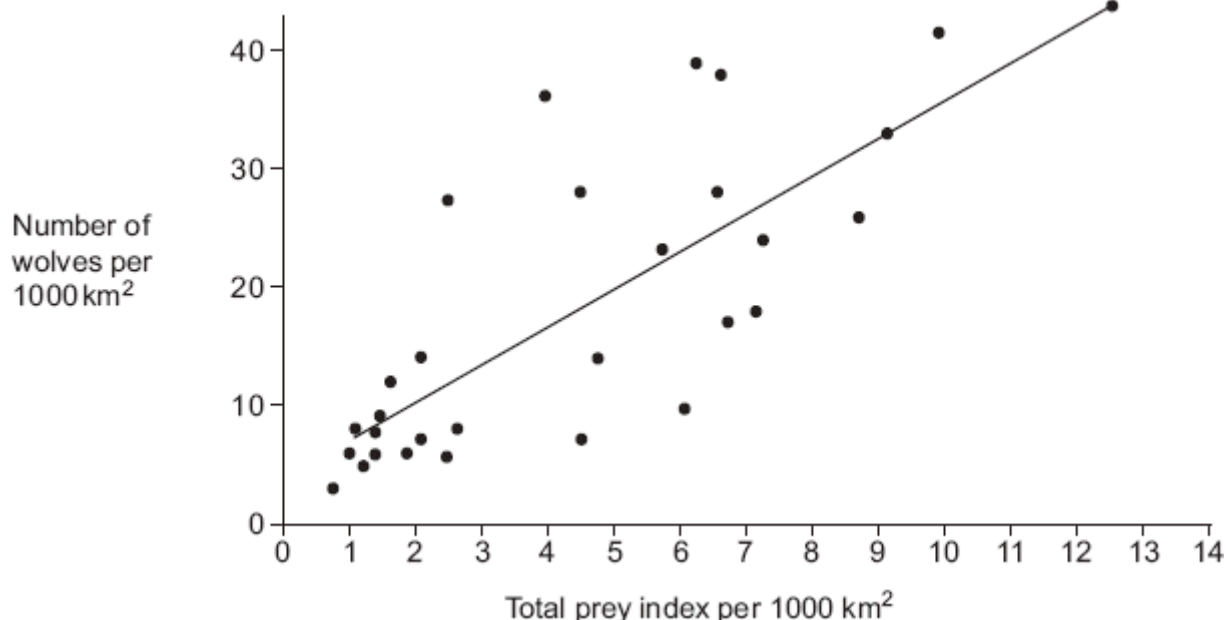
(i) Explain why DNA on the Y chromosome helped them to reach this conclusion.

(1)

(ii) Suggest why DNA in the mitochondria helped them to reach this conclusion.

(1)

Wolves eat different mammals. An ecologist investigated factors that affect wolf numbers in North America. He collected data from different field studies carried out in different places. The graph shows his results.



- (d) (i) The wolf numbers are given per unit area. Explain why.

(2)

- (ii) The ecologist calculated the total prey index for each of the places that had been studied. In order to do this, he gave each prey species a value based on how much food was available to wolves from the prey animal concerned. He called this value the prey index.

The ecologist considered that the prey index gave a better idea of the food available than the prey biomass in kg. Suggest why the prey index gives a better idea of food available.

(2)

- (e) The ecologist calculated the total prey index by combining the prey indices and the total number of animals of each species present in 1000 km². He plotted this information on the graph. What does the graph suggest about the factors that determine wolf numbers in North America? Explain your answer.

(2)
(Total 12 marks)

Q8.

- (a) What is meant by a gene?

(2)

The polymerase chain reaction (PCR) can be used to obtain many copies of a particular gene.

- (b) Explain how the strands of DNA are separated during the PCR.

(2)

- (c) In a particular PCR, two different primers are added to the DNA.

- (i) Why are primers required?

(1)

- (ii) Suggest why two different primers are required.

(1)

- (d) Starting with a single molecule of DNA, the polymerase chain reaction was allowed to go through three complete cycles. How many molecules of DNA would be produced?

Answer _____
(1)

Q9.

Read the following passage.

The giant panda is one of the rarest animals in the world and is considered to be on the brink of extinction in the wild. Giant pandas have been kept and bred in zoos with the hope that they could be released into the wild. One worry is that small populations, like those in zoos, reduce the genetic variation needed to allow the species to adapt to changing conditions. Unfortunately, pandas find it difficult to reproduce in captivity. Fertilisation of the females is guaranteed only by insemination with semen from several males. With so many potential fathers, the true paternity of the cubs is not clear. It is important to identify the fathers to maintain genetic variation.

5

10

Panda faeces can be collected in the wild. The faeces contain DNA from the panda, from the bamboo on which they feed and from bacteria. The DNA is subjected to the polymerase chain reaction (PCR). The primers used attach only to the panda DNA. The resulting DNA is subjected to genetic fingerprinting. This can help us to count the number of individuals in the wild because it allows us to identify individual pandas.

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

- (a) Describe how genetic fingerprinting may be carried out on a sample of panda DNA.

(6)

- (b) (i) Explain how genetic fingerprinting allows scientists to identify the father of a

particular panda cub.

(2)

- (ii) When pandas are bred in zoos, it is important to ensure only unrelated pandas breed. Suggest how genetic fingerprints might be used to do this.

(1)

- (c) (i) Suggest why panda DNA is found in faeces. (line 10)

(1)

- (ii) Explain why the PCR is carried out on the DNA from the faeces. (line 12)

(1)

- (iii) Explain why the primers used in the PCR will bind to panda DNA, but not to DNA from bacteria or bamboo. (line 12)

(2)

- (d) DNA from wild pandas could also be obtained from blood samples. Suggest **two** advantages of using faeces, rather than blood samples, to obtain DNA from pandas.

1. _____

2. _____

(2)

(Total 15 marks)

Q10.

Read the following passage.

DNA tests were used to confirm the identity of deposed Iraqi leader Saddam Hussein, after his capture in December 2003. DNA tests were carried out to prove the suspect was not one of the many alleged “look alikes” of the former leader.

5 Firstly, the DNA was extracted from the mouth of the captured man using a swab. Great care was taken to check that the swab did not become contaminated with any other DNA. DNA extracted from the swab was then subjected to a standard technique called the polymerase chain reaction (PCR), which takes a couple of hours. Lastly, the sample was “typed” to give the genetic fingerprint. This was produced within 24 hours of Saddam Hussein’s capture. Tests for use in criminal cases often take much longer because samples are very small or contaminated.

It appears that Hussein's genetic fingerprint was already stored away for comparison. This was obtained from personal items such as his toothbrush. DNA from the toothbrush would have been subjected to PCR before a DNA fingerprint could have been obtained.

Source: adapted from SHAONI BHATTACHARYA, *New Scientist* 15 December, 2003

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

- (a) Describe how the technique of genetic fingerprinting is carried out and explain how it can be used to identify a person, such as Saddam Hussein.

[illegible]

(6)

- (b) Explain how DNA could be present on a toothbrush (line 12).

(2)

- (c) (i) Explain why the polymerase chain reaction was used on the sample of DNA from the toothbrush (lines 12-13).

(2)

- (ii) Explain **one** way in which the polymerase chain reaction differs from DNA replication in a cell.

(2)

- (d) Tests for use in criminal cases often take much longer because samples are very small or contaminated (lines 8-10). Explain why it takes longer to obtain a genetic fingerprint if the sample is

- (i) very small;

(1)

- (ii) contaminated.

(2)

(Total 15 marks)

Q11.

A gene was broken into fragments using enzyme **Z**. The mixture of fragments produced was then separated by electrophoresis.

(a) What type of enzyme is enzyme **Z**?

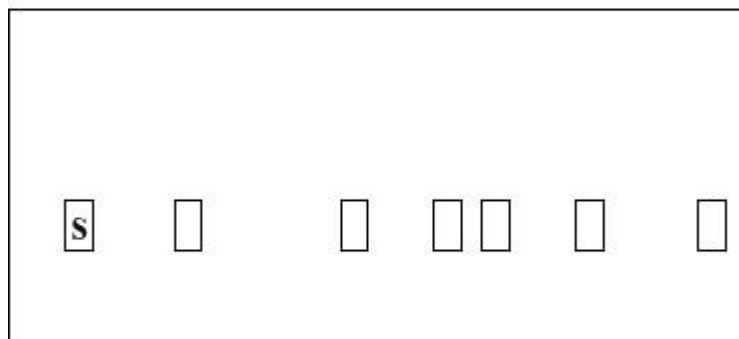
(1)

The table shows the number of base pairs present in the fragments.

Fragment	Number of base pairs ($\times 10^3$)
1	4.65
2	5.72
3	10.71
4	2.39
5	5.35
6	7.53

The diagram shows the electrophoresis gel used. The mixture of fragments was placed at the start point marked **S** and the process started. The boxes indicate the positions reached by the different fragments.

Direction of movement of fragments



(b) Explain why base pairs are a suitable way of measuring the length of a piece of DNA.

(2)

(c) (i) Write **6** above the appropriate box on the diagram to show the position you would expect fragment **6** to have reached.

(1)

- (ii) Explain how you arrived at your answer.

(1)

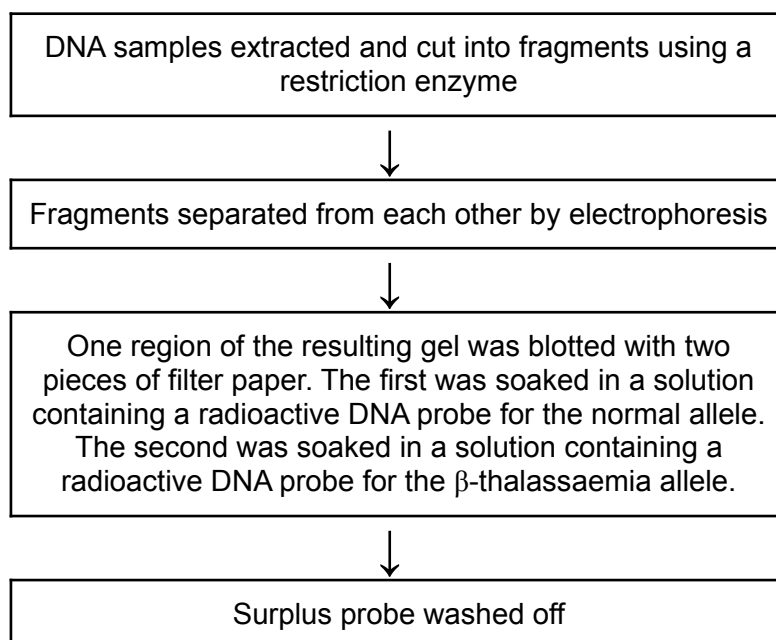
- (d) Enzyme **Z** recognises a particular sequence of bases in the gene. How many times does this sequence appear in the DNA of this gene?

(1)

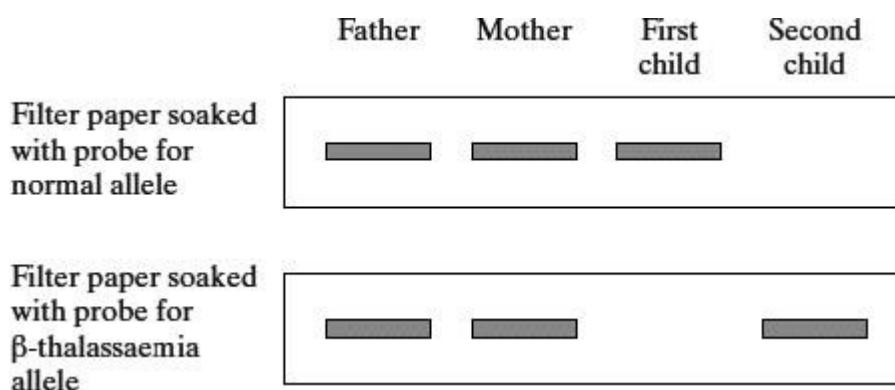
(Total 6 marks)

Q12.

β -thalassaemia is a genetic condition in which abnormal haemoglobin is produced. In one form, the recessive allele for β -thalassaemia, **t**, differs from the normal allele, **T**, by a single base-pair. A radioactive DNA probe was used to investigate the genotypes of four members of one family. The flowchart summarises the technique involved.



The diagram below shows the appearance of the two pieces of filter paper which resulted from the investigation.



- (a) What is the probability that the next child that this couple have is a girl who has β -

thalassaemia? Explain your answer.

(3)

- (b) (i) The fragment of DNA containing the normal allele and the fragment with the β -thalassaemia allele moved the same distance on the gel. Explain why.

(2)

- (ii) The allele for β -thalassaemia differs from the normal allele by only one base-pair. Explain why the probe used to identify these alleles consists of a piece of DNA twenty bases in length and not just one base.

(2)

(Total 7 marks)

Q13.

Read the following passage.

Shark-fin soup is an expensive delicacy. To provide the basic ingredient, fishermen catch the sharks, hack the fins off and throw the dead bodies back into the ocean. But sharks are slow to mature and produce only a few offspring at a time, so they are vulnerable to overfishing. Monitoring the shark-fin trade is difficult, as once a fin has been cut off, it can be extremely difficult to work out precisely from which species it was taken.

5

The DNA from different species of sharks shows some differences in base sequence. This has enabled a new genetic fingerprinting technique to be developed. This technique would allow conservationists and fisheries managers to assess which of the 400 shark species are most threatened by the trade in shark fins.

10 An identification process has been developed using a range of “primers”. These are short pieces of single-stranded DNA that are complementary to a particular sequence of DNA.

Each primer is specific to the DNA of one shark species.

- 15 The primers are added to DNA taken from a shark's fin and the polymerase chain reaction is carried out. Only two primers, one at each end of a certain piece of DNA, will bind. The piece of DNA between the primers is replicated by the polymerase chain reaction. The primers that bind are specific to a particular species of shark and the length of the DNA fragment replicated differs for each species. When this DNA is run in an electrophoresis gel it produces a single band, enabling the researchers to identify which species of shark is involved.

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

- (a) (i) Explain why the DNA for each species of shark shows differences in base sequence (line 6).

(2)

- (ii) Each primer is specific to the DNA of one shark species (line 12).

Explain why a particular primer will only bind to the DNA of one species.

(2)

- (iii) The length of the replicated DNA fragment is different for each species.

Explain why this is important in identifying the shark species involved.

(3)

- (b) In conventional DNA fingerprinting, a series of bands is produced on the electrophoresis gel, resembling the rungs of a ladder. When the DNA in this new genetic fingerprinting technique is run in an electrophoresis gel it produces just one of these 'rungs'.

(c) Describe the polymerase chain reaction.

[illegible]

(Total 15 marks)

Read the following passage.

The chart shows some of the results obtained from the genetic fingerprinting of seven of the skeletons, three children and four adults.

Child 1	Child 2	Child 3	Adult 1	Adult 2	Adult 3	Adult 4
		—			—	
—	—				—	—
—	—	—	—	—	—	—
—	—	—		—	—	
—		—	—	—		—
—	—	—	—	—		—
—	—	—	—	—	—	
—	—	—	—	—	—	
—	—	—	—	—	—	
—	—	—	—	—	—	
				—		—
—		—	—		—	
	—	—			—	

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

- (a) Explain why the polymerase chain reaction was used in this investigation.

(2)

- (b) In the polymerase chain reaction, DNA is heated to 95 °C and nucleotides, enzymes and DNA primers are added to the mixture.

- (i) Explain why the DNA is heated to 95 °C.

(1)

- (ii) What are DNA *primers*?

(1)

[illegible]

(2)
(Total 15 marks)

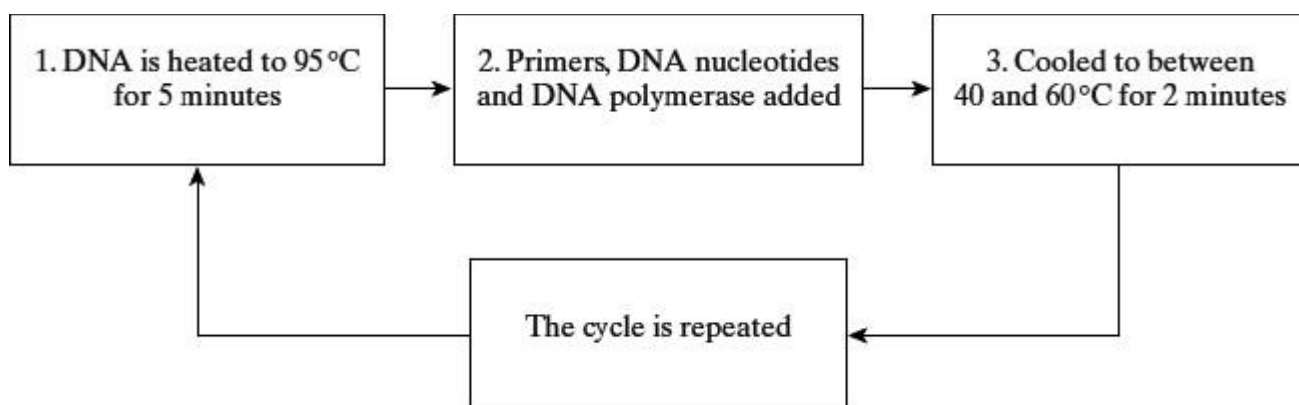
Q15.

The polymerase chain reaction (PCR) can be used to produce large quantities of DNA. Describe how the PCR is carried out.

(Total 6 marks)

Q16.

The polymerase chain reaction is a process which can be carried out in a laboratory to replicate DNA. The diagram shows the main stages involved in the polymerase chain reaction.



(a) Explain why DNA is heated to 95 °C.

-
- (1)
- (b) What is the role of
- (i) a primer in this process;
- _____
- _____
- (1)
- (ii) DNA polymerase?
- _____
- _____
- (1)
- (c) (i) How many DNA molecules will have been produced from one molecule of DNA after 6 complete cycles?
- _____
- (1)
- (ii) Suggest **one** use of the polymerase chain reaction.
- _____
- _____
- (1)
- (d) Give **two** ways in which the polymerase chain reaction differs from the process of transcription.
1. _____
- _____
2. _____
- _____
- (2)
- (Total 7 marks)

Q17.

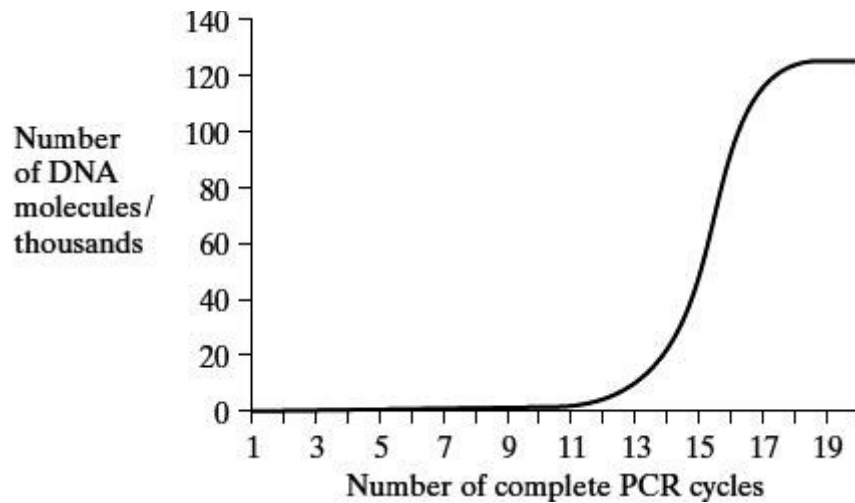
- (a) Explain the reason for each of the following in the polymerase chain reaction (PCR).
- (i) DNA is heated to 95 °C.
- _____
- _____
- (1)
- (ii) DNA polymerase used is heat-stable.

(1)

- (iii) The reaction mixture is cooled to 40 °C.

(1)

- (b) The graph shows the number of DNA molecules made using PCR, starting with one molecule.



- (i) Explain the shape of the curve from cycles 1 to 16.

(2)

- (ii) Suggest **one** explanation for the levelling out of the curve from cycles 17 to 20.

(2)

(Total 7 marks)

Mark schemes

Q1.

- (a) (All) the DNA in a cell/organism;
Accept
 '(all) the 'genes'/alleles' 'genetic material/code' in a cell/organism/ person'
 'the total number of DNA bases in a cell/organism'
Reject all the DNA/ genes within a species
 1
- (b) 1. (Transcriptional factor/antibody) has a specific/tertiary structure/shape;
Accept (antibody) has a specific variable region
Accept (transcription factor/antibody) has a specific binding site
Reject active site but only once.
 2. Complementary (shape/structure);
Reject active site but only once.
 2
- (c) DNA, transcription factor and antibody;
Accept Nucleotides for DNA
Ignore 'reference to chemicals'
 1
- (d) Binds to P34 gene/DNA/mRNA
OR
 Binds to transcription factor gene/DNA
OR
 Binds to promoter;
Reject binds to transcription factor
 1
- (e) 1. Restriction (endonuclease/enzyme) to cut plasmid/vector;
 2. Ligase joins gene/DNA to plasmid/vector;
 2
- (f) 1. Mass/number of amino acids/polypeptides;
Accept weight for mass
Ignore density/size
Accept length of polypeptide/amino acid chain
Accept primary structure /sequence of amino acids.
Accept tertiary structure
 2. Charge;
 3. R groups (differ);

2 max

[9]

Q2.

- (a) 1. Cell membranes made from phospholipid;
Note: candidates may make both mark points in once statement
2. (Detergent) dissolves membranes / phospholipid (bilayer);
Ignore breaks down
Reject hydrolysis
- (b) 1. Spin (liquid / supernatant) at (very) high speed / high g;
*Need context of high, **not** just 'faster', 'higher' in context of use of bench centrifuge*
Accept high centrifugal force
Award 1 max if ref. to organelle separation
2. Molecules / CENP-W separates depending on (molecular) mass / size / density;
Accept weight
- (c) 1. siRNA binds to / destroys mRNA for CENP-W;
Reject if siRNA binds to gene / DNA
*Context is important, siRNA acts on mRNA for CENP-W, **not** tubulin.*
*Ref. to CENP-W required **once** for MP1 and MP2*
2. Prevents translation of CENP-W;
*Context is important, siRNA acts on mRNA for CENP-W, **not** tubulin.*
*Ref. to CENP-W required **once** for MP1 and MP2*
Accept reduces translation of CENP-W
3. (After / as) CENP-W reduces **so** does tubulin production;

2

2

3

[7]

Q3.

- (a) 1. Cut (DNA) at same (base) sequence / (recognition) sequence;
Accept: cut DNA at same place
2. (So) get (fragments with gene) **R** / required gene.
Accept: 'allele' for 'gene' / same gene
- (b) 1. Each has / they have a specific base sequence;
2. That is complementary (to allele r or R).
Accept description of 'complementary'
- (c) 1. Fragments L from parent rr, because all longer fragments / 195 base pair fragments;
Ignore: references to fragments that move further / less,

2

2

require identification of longer / shorter or 195 / 135

Accept: (homozygous) recessive

2. Fragments N from parent RR, because all shorter fragments / 135 base pair fragments;

1 and 2 Accept: A3 for 195 and A4 for 135

2. Accept: (homozygous) dominant

3. (M from) offspring heterozygous / Rr / have both 195 and 135 base pair fragments.

Accept: have both bands / strips

Reject: primer longer / shorter

3

- (d) 1. (Cells in mitosis) chromosomes visible;
2. (So) can see which chromosome DNA probe attached to.

2

- (e) (i) 1. For comparison with resistant flies / other (two) experiments / groups;
Ignore: compare results / data / no other factors
2. To see death rate (in non-resistant) / to see effect of insecticide in non-resistant / normal flies.
Accept: 'pesticide' as 'insecticide'
Accept to see that insecticide worked / to see effect of enzyme

2

- (ii) (PM must be involved because)
1. Few resistant flies die (without inhibitor);
2. More inhibited flies die than resistant flies;
3. (PM) inhibited flies die faster (than resistant flies);
(Other factors must be involved because)
4. Some resistant flies die;
5. But (with inhibitor) still have greater resistance / die slower than non-resistant flies.
Accept: (with inhibitor) die slower than non-resistant flies

4 max

[15]

Q4.

- (a) Reverse transcriptase;
- (b) 1. Probe (base sequence) complementary (to DNA of allele A / where A is (and) binds by forming base pairs / hydrogen bonds;
Accept gene A
2. So (only) this DNA labelled / has green dye / gives out (green) light;
Accept glows for green light
- (c) (i) 1. More probe binding / more cDNA / mRNA / more allele / gene A means more light;

1

2

2. DNA (with **A**) doubles each (PCR) cycle;
3. So light (approximately) doubles / curve steepens more and more (each cycle) / curve goes up exponentially / increases even faster;

3

(ii) (**G** because)

1. (Heterozygous) only has half the amount of probe for **A** attaching / only half the amount of DNA / allele A (to bind to);
Accept only one A to bind to
2. (So,) only produced (about) half the light / glow / intensity (of **H**) (per cycle of PCR);
If reference to 'half' for point 1, allow 'less light' in 2.

2

[8]

Q5.

- (a) (i)
1. Negative correlation;
Accept: description for 'negative correlation'
Neutral: 'correlation'
Reject: positive correlation
 2. Wide range;
 3. Overlap;
 4. (Graph suggests that) other factors may be involved (in age of onset);
2 / 3 Accept the use of figures from the graph
2 / 3 Can refer to age of onset or number of CAG repeats
Ignore references to methodology

3 max

- (ii)
1. Age of onset can be high / symptoms appear later in life;
Accept: 'gene' for 'allele'
 2. (So) individuals have already had children / allele has been passed on;

OR

3. Individuals have passed on the allele / already had children;
4. Before symptoms occur;

2 max

- (b) (i)
1. Person **K**;
 2. (As has) high(est) band / band that travelled a short(est) distance / (er) so has large(st) fragment / number of CAG repeats;
Must correctly link distance moved and fragment size

2

- (ii) Run fragments of known length / CAG repeats (at the same time);
Accept: references to a DNA ladder / DNA markers
Do not accept DNA sequencing
- (iii) Homozygous / (CAG) fragments are the same length / size / mass;
Accept: small fragment has run off gel / travelled further

1

1

[9]

Q6.

21 – 25	Extended abstract Generalised beyond specific context	Response shows holistic approach to the question with a fully integrated answer which makes clear links between several different topics and the theme of the question. Biology is detailed and comprehensive A-level content, uses appropriate terminology, and is very well written and always clearly explained. No significant errors or irrelevant material. For top marks in the band, the answer shows evidence of reading beyond specification requirements.
16 – 20	Relational Integrated into a whole	Response links several topics to the main theme of the question, to form a series of interrelated points which are clearly explained. Biology is fundamentally correct A-level content and contains some points which are detailed, though there may be some which are less well developed, with appropriate use of terminology. Perhaps one significant error and, or, one irrelevant topic which detracts from the overall quality of the answer.
11 – 15	Multistructural Several aspects covered but they are unrelated	Response mostly deals with suitable topics but they are not interrelated and links are not made to the theme of the question. Biology is usually correct A-level content, though it lacks detail. It is usually clearly explained and generally uses appropriate terminology. Some significant errors and, or, more than one irrelevant topic.
6 – 10	Unistructural Only one or few aspects covered	Response predominantly deals with only one or two topics that relate to the question. Biology presented shows some superficial A-level content that may be poorly explained, lacking in detail,

		or show limited use of appropriate terminology. May contain a number of significant errors and, or, irrelevant topics.
1 – 5	Unfocused	Response only indirectly addresses the theme of the question and merely presents a series of biological facts which are usually descriptive in nature or poorly explained and at times may be factually incorrect. Content and terminology is generally below A-level. May contain a large number of errors and, or, irrelevant topics.
0		Nothing of relevance or no response.

Commentary on terms and statements in the levels mark scheme

The levels mark scheme for the essay contains a number of words and statements that are open to different interpretations. This commentary defines the meanings of these words and statements in the context of marking the essay. Many words and statements are used in the descriptions of more than one level of response. The definitions of these remain the same throughout.

Levels mark scheme word/statement	Definition
Holistic	Synoptic, drawing from different topics (usually sections of the specification)
A fully integrated answer which makes clear links between several different topics and the theme of the question	All topics relate to the title and theme of the essay; for example, explaining the biological importance of a process. When considering, for example, the importance of a process, the explanation must be at A-level standard. ‘Several’ here is defined as at least four topic areas from the specification covered. This means some sentences, not just a word or two. It does not mean using many examples from one topic area.
Biology is detailed and comprehensive A-level content, uses appropriate terminology, and is very well written and always clearly explained.	Detailed and comprehensive A-level content is the specification content. Terminology is that used in the specification. Well written and clearly explained refers mainly to biological content and use of terminology. Prose, handwriting and spelling are secondary

	considerations. Phonetic spelling is accepted, unless examiners are instructed not to do so for particular words; for example, glucagon, glucose and glycogen.
No significant errors or irrelevant material.	A significant error is one which significantly detracts from the biological accuracy or correctness of a described example. This will usually involve more than one word. Irrelevant material is several lines (or more) that clearly fails to address the title, or the theme of the title.
For top marks in the band, the answer shows evidence of reading beyond specification requirements.	An example that is relevant to the title and is not required in the specification content. The example must be used at A-level standard.
Response mostly deals with suitable topics but they are not interrelated and links are not made to the theme of the question.	Not addressing the biological theme of the essay (e.g. importance) at <u>A-level standard</u> .

Please note that to obtain full credit, students must use information to show **the importance of Using DNA in science and technology**.

Topics

DNA and classification	
2.2	Structure of DNA
2.3	Differences in DNA lead to genetic diversity
2.9	Comparison of DNA base sequences
Genetic engineering and making useful substances	
2.5	Plasmids
5.8	The use of recombinant DNA to produce transformed organisms that benefit humans
Other uses of DNA	
2.5	Cell cycle and treatment of cancer
5.8	Gene therapy; Medical diagnosis and the treatment of human disease; The use of DNA probes to screen patients for clinically important genes.

In order to fully address the question and reach the highest mark bands students must also include at least four topics in their answer, to demonstrate a synoptic approach to the

essay.

Students may be able to show the relevance of other topics from the specification.

Note, other topics from beyond the specification can be used, providing they relate to the title and contain factually correct material of at least an A-level standard. Credit should not be given for topics beyond the specification which are below A-level standard.

[25]

Q7.

- (a) (i) To cut the DNA;
Reject breakdown, cutting out 1
- (ii) To separate the (pieces of) DNA; 1
- (b) Complimentary base sequence / complementary DNA; binds to both (haplotypes);
Label would show up in both;
Idea of complimentarity required 2
- (c) (i) Y chromosome inherited / comes from male parents / only found in males; 1
- (ii) Mitochondria in egg / female gamete / no mitochondria come from sperm / male gamete; 1
- (d) (i) Allows comparison;
Different (sized) areas covered; 2
- (ii) Wolves do not eat all of prey animal / do not eat (large) bones / skin;
Inedible parts make up different proportions / wolf eats different proportions; 2
- (e) Limited by food / prey; as prey increases so do wolf numbers / positive correlation;
Large range so other factors involved; 2

[12]

Q8.

- (a) a length of DNA;
that codes for a single protein / polypeptide; 2
- (b) by heating;

- to break the H-bonds (between complementary bases); 2
- (c) (i) to allow the DNA polymerase to attach / start addition of nucleotides / mark start and end of sequence to be copied / prevents strands re-joining; 1
- (ii) because the sequences at the ends of the target sequence are different / one is at the beginning and one at the end; 1
- (d) 8; 1
- accept 7*

[7]

Q9.

- (a) 1 DNA is cut;
- 2 Using restriction enzyme;
- 3 Use electrophoresis;
- 4 Separates according to length / mass;
- 5 Southern blotting / transfer to (nylon) membrane;
- 6 Make single-stranded;
- 7 Apply probe;
- 8 Radioactive / fluorescent;
- 9 Reference to tandem repeats / VNTRs / minisatellites;
- 10 Autoradiography / eq;
- 8 and 10 should be consistent
- max 6
- (b) (i) All bands in cub which don't come from mother;
- Must be in father's DNA fingerprint;
- Principle that all bands in cub must come from mother and father = 1*
- 2
- (ii) Select pairs with dissimilar DNA fingerprints; 1
- (c) (i) Cells (from panda) in faeces / gut cells / blood cells; 1
- (ii) To increase amount of DNA / only small amount present; 1
- (iii) DNA / primer has specific base-sequence;
- Reference to specific / complementary base-pairing;

- (d) Taking samples from animals causes stress / injury to animal;

Difficult to find animals;

Pandas are dangerous / threat to human;

max 2

[15]

Q10.

- (a) 1. DNA is cut;
2. using restriction enzyme;
3. electrophoresis;
4. separates according to length / mass / size;
5. DNA made single-stranded;
6. transfer to membrane / Southern blotting;
7. apply probe;
8. radioactive / single stranded / detected on film / fluorescent;
9. reference to tandem repeats / VNTRs / minisatellites;
10. pattern unique to every individual;

6 max

- (b) cells on toothbrush;
DNA present in cell;

2

- (c) (i) toothbrush gives small sample of DNA / need more DNA for analysis;
PCR gives many copies;

2

- (ii) uses heat;
to separate strands;
OR
PCR replicates pieces of DNA;
because DNA has been cut;
OR
primer added in PCR;
to initiate replication

2 max

- (d) (i) PCR / amplification needed;

1

- (ii) other DNA present; need to identify 'required' DNA from rest;

2

[15]

Q11.

- (a) Endonuclease / restriction enzyme;

1

- (b) DNA made of base pairs;
Each base pair is same length / occupies same distance along backbone;

2

- | | | | | |
|-----|------|---|---|-----|
| (c) | (i) | Second blank box from left labelled 6; | 1 | |
| | (ii) | Distance moved depends on length / number of base pairs /
second longest fragment / second shortest distance identified; | 1 | |
| (d) | | 5; | 1 | [6] |

Q12.

- | | | | | |
|-----|------|---|---|-----|
| (a) | | Mother and father both heterozygotes / Tt / carriers;
Probability of thalassaemia 1/4 and female 1/2;
Probability of both 1/8; | 3 | |
| (b) | (i) | Cut at same base sequence as same enzyme used;
Fragments are same length / size / have same charge; | 2 | |
| | (ii) | Single base occurs many times;
Sequence of 20 unlikely to occur elsewhere;
<i>Allow one mark for establishing the principle where neither marking
point clearly made.</i> | 2 | [7] |

Q13.

- | | | | | |
|-----|-------|--|-------|--|
| (a) | (i) | Different genes / characteristics / features;
Reference to mutations;
Or
Base sequence determines protein;
Different species have different protein sequences; | max 2 | |
| | (ii) | Primer has different DNA sequence;
DNA specific / complementary base-pairing; | 2 | |
| | (iii) | Electrophoresis separates DNA;
(So they can be) identified by position on gel;
Smaller / shortest fragments travel furthest / quicker / or
reverse argument; | 3 | |
| (b) | | (<i>conventional</i>) Many lengths / all DNA / (<i>new</i>) one length;
Each rung is DNA of one / specific length; | 2 | |
| (c) | | 1 Heat DNA;
2 Breaks hydrogen bonds / separates strands;
3 Add primers;
4 Add nucleotides;
5 Cool;
6 (to allow) binding of nucleotides / primers;
7 <u>DNA</u> polymerase; | | |

- 8 Role of (DNA) polymerase;
9 Repeat cycle many times;

max 6

[15]

Q14.

- (a) only small amounts obtained / PCR increases the amount / mass of DNA;
so enough DNA available for genetic fingerprinting;

2

- (b) (i) to separate the two strands of the DNA /
to break the hydrogen bonds;
(Reject "unzip")

1

- (ii) short lengths / fragments of DNA / nucleotides /
single stranded DNA;

1

- (iii) to mark beginning and / or ends of the part of DNA needed /
for attachment of enzymes or nucleotides / initiator /
keeps strands apart;

1

- (iv) would not be denatured;
must be heated to 95 °C / must withstand high temps;

2

- (c) 1 DNA extracted from sample;
2 DNA cut / hydrolysed into segments using restriction endonucleases;
3 must leave minisatellites / required core sequences intact;
4 DNA fragments separated using electrophoresis;
5 detail of process e.g. mixture put into wells on gel and electric
current passed through;
6 immerse gel in alkaline solution / two strands of DNA separated;
7 Southern blotting / cover with nylon / absorbent paper (to absorb DNA);
8 DNA fixed to nylon / membrane using uv light
9 radioactive marker / probe added (which is picked up by required
fragments) / complementary to minisatellites;
10 (areas with probe) identified using X-ray film / autoradiography;

max 6

- (d) adult 3;
this is only one which, (with number 1), can provide (all) the DNA
fragments which children have / all bars match;
(Reject 'genes')

2

[15]

Q15.

- 1 DNA heated to 90 to 95°C;
2 strands separate;
3 cooled / to temperature below 70°C
4 primers bind;
5 nucleotides attach;
6 by complementary base pairing;

- 7 temperature 70 - 75°C;
- 8 DNA polymerase joins nucleotides together;
- 9 cycle repeated;

6 max

[6]

Q16.

- (a) to separate the two strands / break hydrogen bonds; 1
- (b) (i) enables replication / sequencing to start (*allow keeps strands separate*); 1
- (ii) joins DNA nucleotides (*not complementary bases*); 1
- (c) (i) 64; 1
- (ii) replication of DNA from crime scene / tissue sample / for DNA sequencing / gene cloning; 1
- (d) (transcription uses) RNA polymerase; RNA nucleotides / uracil; one (template) strand / PCR both strands; start / stop codons; (*accept enzyme separates strands*)

2 max

[7]

Q17.

- (a) (i) to separate polynucleotide strands / form single strands; 1
- (ii) not denatured (at 95°C); 1
- (iii) for binding of primers / nucleotides (to DNA strands); 1
- (b) (i) doubling (of DNA) each cycle; but very low numbers to start with, so appears flat then exponential growth; 2
- (ii) suggestion; with explanation e.g.:
 - nucleotides being used up;
 - so less / nothing to make complementary chains;
 - primers used up;
 - so cannot start complementary chains;
 - enzymes losing activity / denatured;
 - so no polymerisation of complementary strands;

Examiner reports

Q1.

- (a) The mark scheme for this question reflected the considerable variation in acceptable definitions for the term genome. Despite this, nearly 40% of students did not obtain the mark. There some very detailed definitions which often included the DNA in mitochondria and chloroplasts. The most common incorrect responses referred to “all the genes in a chromosome” or “all the genes in a species”.
- (b) Almost 80% of students obtained at least one mark for this question, often for referring to complementary structures. Almost half of these students gained a second mark by referring to a specific tertiary structure, shape or binding site. Weaker responses suggested that the term complementary is equivalent to ‘similar’ or referred to ‘active site’ rather than binding site.
- (c) This proved more difficult than expected with only 26% of students obtaining the mark. Incorrect responses included (unbound) DNA fragments, nucleotides, exons, introns, DNA without an antibody or transcription factor bound to it, and any combination of two of the three parts required to gain the mark. The constituent parts of a DNA molecule were also listed.
- (d) Again, relatively few students (30%) obtained the mark for this question. The most frequent correct responses referred to cDNA binding to the *P34* gene, preventing its transcription, or binding to mRNA, preventing its translation. The binding of the cDNA to mRNA, resulting in the destruction of mRNA, was also described. Binding to the promoter region was less frequently awarded. A significant minority of students mentioned that cDNA prevented RNA polymerase binding to the *P34* gene or just prevented its transcription, but with no suggestion of how. Incorrect responses often referred to cDNA increasing/decreasing methylation of the *P34* gene or simply that cDNA could not be transcribed into mRNA and translated to form the *P34* protein.
- (e) Considering that this question was relatively straightforward, it proved to be a very effective discriminator. Surprisingly, many students could not name both enzymes correctly. Consequently, only 35% of students obtained both marks for this question and 28% obtained a single mark. DNA helicase, reverse transcriptase and DNA polymerase were frequent incorrect responses. Even when the correct enzymes were named, students did not always describe the precise roles of the enzymes in the formation of recombinant plasmids. These descriptions were often too vague, e.g. “produces sticky ends” or “joins DNA together”, without any mention of the plasmid/vector. A significant minority referred to incorrect types of bonds when outlining the role of correctly named enzymes.
- (f) A significant number of students had difficulty applying their knowledge of electrophoresis to separating proteins. Only 14% of students gained both marks for this question, however 50% did at least obtain a single mark. Many students simply reverted to discussing DNA, especially VNTRs, or referred to DNA and protein, often suggesting that proteins consist of bases. Common responses which were not credited often referred to ‘size’ and ‘mass’, with no reference to the structure of proteins. However, better answers did obtain a mark when mass/size was linked to the number of amino acids or polypeptides. Difference in charge was the most frequently awarded marking point. Far fewer students mentioned different ‘R groups’ or ‘variable groups’.

Q2.

Questions (a) and (b) required students to evaluate methods used by scientists (AO3), on the basis of information provided and their own knowledge. In question (a), 38% of students correctly noted that cell membranes have a phospholipid bilayer or are made (mainly) from phospholipid, and then suggested that the detergent would dissolve the phospholipids (in water). Some 30% obtained one mark for the point about phospholipids but then wrote about breaking down the membrane, or even hydrolysing it. Students were expected to use the information given and pick up on the fact that the detergent dissolves lipids (in water). The question discriminated well.

In question (b), it was apparent that most students are not familiar with ultracentrifugation as a method for separating molecules. Nearly 60% did obtain one mark, for stating that CENP-W would separate on the basis of its mass/size/density/weight during centrifugation. Only 7% clearly expressed the idea that separating *molecules* requires centrifugation at very high speeds. Many students wrote apparently rote-learned answers about cell fractionation and separating organelles; many referred to CENP-W as an organelle. There were some really good answers in which students wrote about removing organelles by spinning at low speeds and then separating molecules in the supernatant by spinning at very high speeds.

Question (c) produced the best discrimination on the paper. It was pleasing to see that many students had at least some understanding of RNA interference. About 17% of students obtained all three marks. They clearly stated that the siRNA binds to mRNA for CENP-W and this prevents the mRNA from being translated. They then linked this to a reduction in CENP-W and a resulting reduction in tubulin production. About 16% obtained two marks, usually for a reduction in translation and the subsequent reduction in CENP-W and then tubulin. Many of these students wrongly thought that siRNA would prevent transcription, with a lack of mRNA leading to reduced translation. Students who failed to score (37%) were very confused about what siRNA binds to and where. They were often unclear, or wrong, with the contexts for CENP-W and tubulin and the reasons for their reductions. A few students gave apparently rote-learned accounts of how siRNA works, with no references to CENP-W (or tubulin), and failed to score.

Q3.

- (a) Over half of students managed to communicate the idea that the same restriction enzymes cut at the same place / recognition sequence on DNA. Only just over 10 percent considered the context of the question and went on to say that this would give fragments containing the same gene (R).
- (b) About half of students obtained both marks in this part and over 40 percent obtained 1. Most managed to convey the idea that there was binding between complementary base sequences on a primer and DNA from an allele. The better answers conveyed the idea of a specific base sequence for each primer.
- (c) Some very good and clear answers were seen to this part and a third of students obtained 3 marks. These students identified the genotypes of L, M and N and explained how they identified them on the basis of the sizes of primer attached to each and how far the bands moved. Quite a large number went on at length about the offspring as represented by M but failed to identify L and N. A few thought that there were two types of offspring because there were two bands for M; they clearly did not understand the simple genetics of the cross.
- (d) This proved far more challenging than expected. The examiners were looking for the idea that chromosomes would be visible (as separate structures) and thus the scientists could see to which chromosome the probe was attached. Fewer than 10 percent got both marks.

- (e) (i) There were quite a large number of attempts at generic 'How Science Works' answers to this part. 50 percent of students obtained 1 mark, usually for suggesting that the control was to see the effect of the insecticide or that it was for comparison with resistant flies. However, only around 10 percent gave both of these.
- (ii) Most students focused on one aspect of the results and scored 2 marks. They frequently spotted that if the enzyme was the only factor in resistance, then the results for the controls and the resistant flies with the inhibitor would be the same. Others focused on evidence for the statement by comparing the resistant flies with resistant flies with the inhibitor added.

Q4.

- (a) About 80% of students identified the enzyme in this part as reverse transcriptase. The commonest wrong answer appeared to be restriction endonuclease.
- (b) Most students obtained one mark for stating that the DNA probe has a base sequence complementary to the DNA of allele A. A third obtained a second mark by going on to state that this allowed it to bind to the target DNA by base pairing, or that this meant only target DNA gives off green light.
- (c) (i) About half of students obtained one mark in this part for noting that the more probe binding to allele A, the more green light there would be. Nearly a quarter obtained a second mark, usually for also noting that the light curve goes up exponentially (or described). Only a few, 14%, obtained all three marks. These students explained that this was because the amount of DNA doubles (approximately) with each PCR cycle.
- (ii) Answers to this part often made references to G being heterozygous because this person had *fewer* A alleles and thus *less* light was produced. The examiners were looking for more precise statements relating to half the amount of A with probe attached and half (approximately) the light produced (at any given time). A third of students obtained both marks.

Q5.

- (a) (i) Although the negative correlation was usually indicated, only the better students appreciated the wide range in the age of onset or the overlap in values. Very few students were aware that the wide range in age of onset for the same number of CAG repeats suggests that other factors may also be involved.
- (ii) A minority of weaker students gave answers that were out of context. Some thought that the allele for Huntington's disease is recessive and would therefore be passed on to offspring without a person knowing. Others thought that people with Huntington's disease would survive well into adulthood and then reproduce.
- (b) (i) Most students correctly identified Person **K** as testing positive for Huntington's disease. They went on to explain that this person has the fragment that moved the shortest distance and linked this to a greater number of CAG repeats. A minority of students failed to link correctly the distance moved with the length of the fragment.
- (ii) Students who failed to gain credit often referred to using DNA sequencing or probes to highlight sequences and 'restriction mapping'.

- (iii) Misconceptions seen in responses by weaker students included partial digestion of DNA, Person L only having one allele of this gene and the probe not being able to attach to the other fragment.

Q6.

Using DNA in science and technology

The very best essays from candidates who selected this option were outstanding. They reviewed, often in great detail, the relevant aspects of the specification although not always incorporating the role of DNA in the classification of organisms. Considering that much of the content of this essay could be drawn from this unit, it was surprising how poor many answers were. Understanding of techniques was often extremely limited, particularly *in vivo* gene cloning and the use of markers. Many essays presented no more than a broad overview either emphasising ethical issues at the expense of biological detail or failing to distinguish established practice from wishful thinking.

Q7.

- (a) Most candidates had some understanding of the function of restriction endonuclease but were not always sure of its role in the investigation described. Thus, there were numerous references to the enzyme “cutting out” particular sections of DNA, these pieces ranging from haplotypes, to genes and even chromosomes. Most candidates correctly suggested that electrophoresis would be involved in separating the DNA fragments, although some were clearly of the opinion that it was the chains of DNA that were separated.
- (b) Candidates were generally able to describe the complementary base sequence present on the probe but seldom progressed to explain how it could be used to show that the haplotypes concerned were the same.
- (c) The majority of candidates linked the Y-chromosome to male inheritance in part (i) although a significant number suggested that the Y-chromosome was inherited from the female. Part (ii) was targeted at stronger candidates, but very few could suggest that mitochondria could only be passed to the offspring in the cytoplasm of the egg.
- (d) The responses to part (i) suggested that while many candidates were aware that giving the units per unit area enabled comparison, they were uncertain as to what was being compared. The most frequent suggestion was that it allowed wolves to be compared with prey numbers. Others wrote about the territorial behaviour of wolves or suggested that the mobility of the animals made counting over a larger area too difficult. In part (ii), better candidates appreciated that wolves ate only part of their prey and that the amount eaten differed with different species of prey.
- (e) Although the positive correlation between prey index and wolf numbers was usually recognised, few progressed to state that this suggested that food must be limiting population size. Unfortunately, the few who pointed out that other factors might possibly be involved rarely linked this conclusion to the spread of data on the graph.

Q8.

- (a) Examiners were very disappointed to read so many incorrect definitions of a gene. The idea that a gene is a ‘strand of DNA’ seems a common misconception. Some indicated that a gene was the entire DNA or that it was part of a chromosome. Few referred to it coding for a polypeptide but instead gave a vague reference to ‘characteristics’ or used examples, e.g., eye colour. A significant number incorrectly believed that the gene was a protein.

- (b) The concept of breaking bonds between the strands was well understood. However some failed to name the bonds correctly. Others incorrectly suggested that the bonds were broken by the action of enzymes (often restriction enzymes) rather than heat. Nevertheless, many candidates scored both marks here.
- (c) A significant proportion of the candidates were unsure of the role of primers, and a larger number could not suggest why two different primers are required.
- (d) Many candidates gave the correct answer of 8. Among the incorrect answers, the commonest were sixteen or six.

Q9.

- (a) Some candidates could not isolate the essential information. Frequently, candidates included the detail about PCR in their responses before embarking on genetic fingerprinting. Maximum marks were regularly scored however.
- (b) Some candidates experienced problems explaining the situation in part (i). Too often they failed to include reference to the maternal DNA in their argument. Most based their argument on matching cub DNA to the paternal DNA, but this would simply show a close relationship. The question wanted candidates to focus on establishing paternity. Many candidates failed to state the obvious in (ii). They regularly indicated that the genetic fingerprinting would be used to find out which pandas were related, without saying that unrelated pandas would have dissimilar bands.
- (c) In (i) a significant number indicated that bamboo DNA would be present, which of course would not be a source of panda DNA. Others suggested that protein, saliva or other body fluids would be a source of DNA. The role of PCR in (ii) was understood by the vast majority. However some did erroneously believe that PCR could be used to separate DNA, or to make the molecule longer. In (iii), all too often the primer was simply described as being specific without reference to the sequence of bases. Many were more familiar with the complementary nature of the primer. Some gave detailed accounts of the role of the primer, without explaining how it could bind to panda DNA but not bacterial or bamboo DNA.
- (d) This part proved challenging, even for the most able. Most candidates appreciated that it would be easier to collect faeces but could not offer a reason. 'Cheaper' 'easier' and 'quicker' were frequent answers, without any details to support the argument. Avoidance of distress to the panda was noted by many. However most were more concerned with the expense of collecting blood. They also discussed at length the fact that red blood cells would not provide any DNA, so they assumed that faeces would be a more concentrated source of DNA.

Q10.

- (a) Most candidates made a reasonable attempt at this part of the question with many scoring maximum marks. However many began their answers at the wrong point and gave details of PCR before describing genetic fingerprinting. The order of the process was also often confused, with many separating the strands of DNA before electrophoresis, or putting in a probe at the start of electrophoresis. Another misconception was the use of X-rays to identify the probe.
- (b) Most realised that cells would be removed from the mouth area when brushing the teeth and many then continued correctly to identify DNA as being in those cells. Few showed a better understanding by referring to the fact that the DNA was in the nucleus. The commonest misconception here was that saliva would contain DNA.

- (c) (i) Many realised that PCR was being used to increase the amount of DNA but failed to link this to the low initial amount of DNA.
- (ii) This was a challenge for all but the best. The most common correct answer as to the difference was that PCR required heating to break the hydrogen bonds. Many candidates were of the opinion that PCR was conservative replication of DNA, while semi-conservative DNA replication occurred in cells.
- (d) (i) Many realised that, with a small sample, it would take time to copy the genetic material.
- (ii) Only the better candidates appreciated that the sample would be contaminated with other DNA, and went on to recognise that it would take time to identify this or remove it. There were many vague answers about contaminants and the need to decontaminate.

Q11.

Unit 2

- (a) Most had no trouble here in identifying endonuclease.
- (b) This was badly done since few mentioned or implied that DNA was made of base pairs and that these occupied the same distance along the backbone. Many made it obvious that they meant the distance from one chain to the other or went on to write about the distances in the electrophoresis gel.
- (c) Although the correct box was often located, the second from the right was almost as commonly given. Appropriate justification for the choice was rewarded in (ii) although many candidates suggested fragment 6 was the fifth rather than the second largest.
- (d) All figures were given here with 5 only rarely chosen, 6 being most common.

Unit 3

In part (a), restriction enzyme was correctly identified by almost all candidates. However, part (b) attracted very poor answers. Few commented that DNA is composed of base pairs, or that all base pairs have the same length. Many simply re-stated the question. In (c) (i), most candidates correctly identified the box, but a significant number identified the penultimate box. These candidates explained in (c)(ii) that the *longest* fragments moved furthest. Although they had the correct idea that DNA separates according to length, they had the relationship the wrong way round. In (d), there were few correct answers. Most gave 6.

Q12.

- (a) Figure 2 showed that the male involved in this cross possessed two alleles and this should have alerted candidates to the fact that the gene concerned was not sex-linked. Most of those who identified it as being autosomal, were able to explain that the probability of the next child having thalassaemia was one in four. Rather less success was enjoyed, however, when it came to combining probabilities, and the mathematics of multiplying fractions was evidently beyond the ability of a number of candidates.
- (b) Part (i) was generally answered well by those candidates who distinguished between the processes of electrophoresis, chromatography and centrifugation. The

basis of separation in this case is the difference in charge and, in DNA, this translates into differences in length. The numerous references to such features as solubility suggested that the distinction between these three processes was not always secure. The difficulties encountered by many in part (ii) stemmed largely from confusion between the terms allele, base and probe. There was, as a result, much inaccurate biology and little opportunity for awarding credit.

Q13.

This question proved to be very demanding for many candidates, although better candidates were able to score all, or almost all, of the available marks. In (a) (i), many candidates recognised that different species of shark have different characteristics, but mostly failed to link these characteristics to proteins or to genes. In (a) (ii), some candidates did realise that each primer had a specific base sequence which would bind to the shark DNA by complementary base-pairing, but weaker candidates simply re-stated the information in the stem. In (a) (iii), many candidates limited their answers to stating that the DNA fragment had to be a different length in each species so it could be identified. How this enabled identification was not stated. On the other hand, good candidates wrote succinctly about electrophoresis separating DNA by size, and explained how the different length fragments would move different distances up the gel. Part (b) was poorly done by most candidates. It was common to find descriptions of genetic fingerprinting here. A few very weak students confused the 'rungs' with base pairs in the DNA molecule. Part (c) was the most accessible part of the question. A large proportion of candidates clearly knew the polymerase chain reaction and gave excellent accounts, often gaining full marks. A minority of very weak candidates confused PCR and transcription.

Q14.

- (a) This yielded good answers backed by common sense.
- (b) Most candidates scored well in part (i). For part (ii) many gave answers which were more suited to part (iii), then failed to offer a relevant answer for (iii). Part (iv) was well done by many.
- (c) Many centres are clearly going into this topic with sufficient depth for their candidates to achieve full credit. The main errors were in confusing the order of events and the exact role of X rays; some discussed a mixture of electrolysis and chromatography and a very few based their answers on ink-based fingerprinting.

Adult 3 was correctly identified in most scripts but some found the reason hard to explain. A discussion of matching 'genes' was a common error.

Q15.

This question gave the candidates the opportunity to display their knowledge of factual material from the specification.

There were some very good answers with many candidates gaining full marks. The events of the cycle were written in the correct sequence and precise details of the temperature regime were well remembered by the majority of candidates. The role of DNA polymerase was often only alluded to, or incorrectly attributed to complementary base pairing. Weaker candidates described either the process of DNA replication in a cell or, less commonly, confused the process with protein synthesis.

Q16.

Although this question produced a wide range of marks, many candidates were able to obtain at least two of the marks available.

- (a) Most candidates gained this mark by referring to separation of the two strands or to the breaking of hydrogen bonds. Vague references to 'uncoiling' or 'unwinding' were not credited.
- (b) Most candidates obtained the mark in part (i) by referring to the role of primers in enabling sequencing to start. However, in part (ii), considerably fewer candidates correctly described the role of DNA polymerase in joining DNA nucleotides together. Many candidates suggested that this enzyme forms the hydrogen bonds between complementary bases.
- (c) Part (i) and part (ii) caused few difficulties for the majority of candidates. In part (i), most candidates correctly calculated that 64 DNA molecules would have been produced after six complete cycles. In part (ii), the use of the polymerase chain reaction was often linked to the amplification of DNA for forensic purposes or for gene cloning.
- (d) This proved to be an effective discriminator with only the best candidates obtaining both marks. Most candidates failed to consider the processes involved and simply referred to the products of PCR and transcription. These answers were not credited. Better candidates often referred to transcription using RNA polymerase, uracil and one template strand.

Q17.

- (a) This question was intended to allow most students to gain marks using their recall of PCR. In the event, few candidates obtained all three marks.
 - (i) Many candidates obtained this mark by explaining that heating to 95°C causes separation of DNA molecules into separate polynucleotide strands. Credit was not given to simple statements restricted to 'breaking' or 'splitting' DNA. A surprising number thought that the heating was to provide the optimum temperature for enzymes.
 - (ii) This was probably the best answered part of the question and many candidates correctly explained that the enzyme is not denatured at the high temperatures used in PCR.
 - (iii) Quite a few candidates wrongly thought that this was the optimum temperature for the enzyme, or the temperature at which the enzyme could start to attach free nucleotides to the complementary bases on single strands. Many correctly identified this as the temperature at which primers or nucleotides can bind (form H bonds) to single-stranded DNA.
- (b)
 - (i) There were many good answers from candidates who understood the concept that the number of DNA molecules doubles with each cycle during PCR but that growth in numbers is slow at first, due to the small number of DNA molecules at the start. Some candidates spoilt their answer by suggesting that there were no DNA molecules at the start.
 - (ii) Many candidates obtained one mark for suggesting that either primers or nucleotides were used up, or that the enzymes lost activity. Quite a few of these candidates were then unable to explain clearly how their suggestion slowed or stopped DNA replication. Some candidates misinterpreted the graph

and saw it as one of rate of enzyme action against time. This led to incorrect suggestions revolving around the idea of the saturation of active sites of enzymes. Credit was given to candidates who suggested that large numbers of DNA molecules produce so many single strands that they re-bind to each other, rather than forming new complementary strands.