Difference in DNA

These practice questions can be used by students and teachers and is

Suitable for AQA A Level 7402 Biology Topic Question

Level: AQA A LEVEL 7402

Subject: Biology

Exam Board: AQA A Level 7402

Topic: Difference in DNA

1

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?

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_____

(1)

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

_____

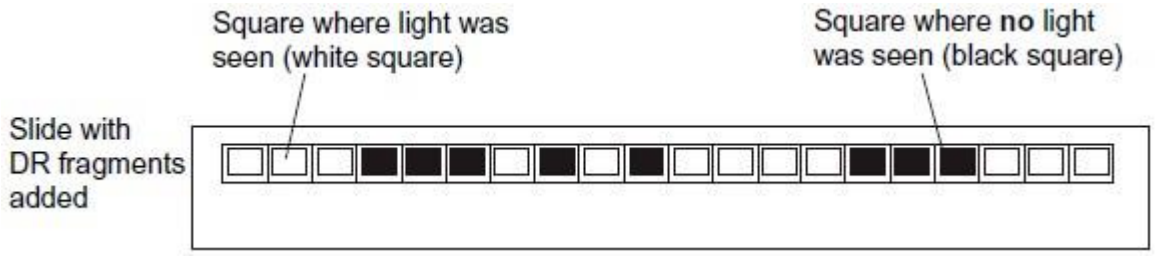
_____

(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 itscomplementary 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)

2 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 DNAfragments.

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

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(2)

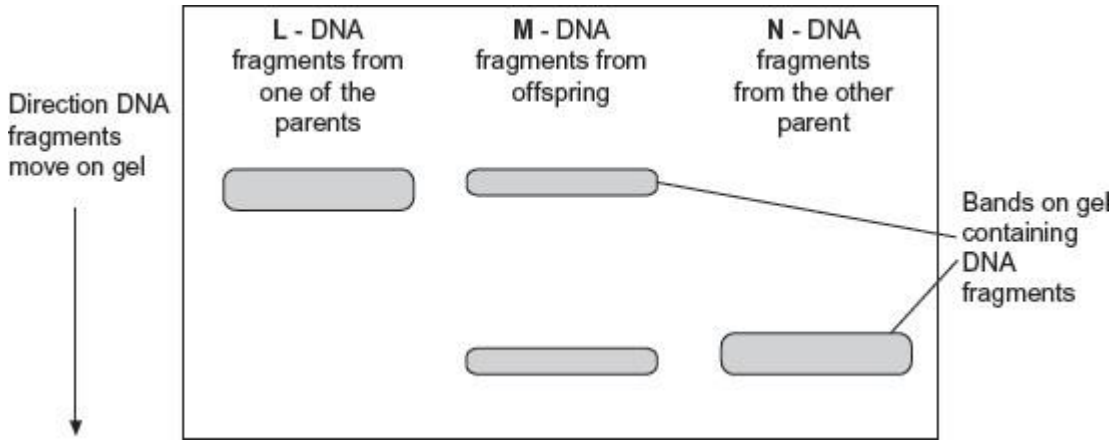
The scientists added two different primers to each sample of DNA fragments for the polymerase chain reaction (PCR).

- 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.

_____

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(2)

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

_____

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[Extra space]

(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 toincreased activity of an enzyme called P450 monooxygenase (PM). This enzyme breaks down insecticides.

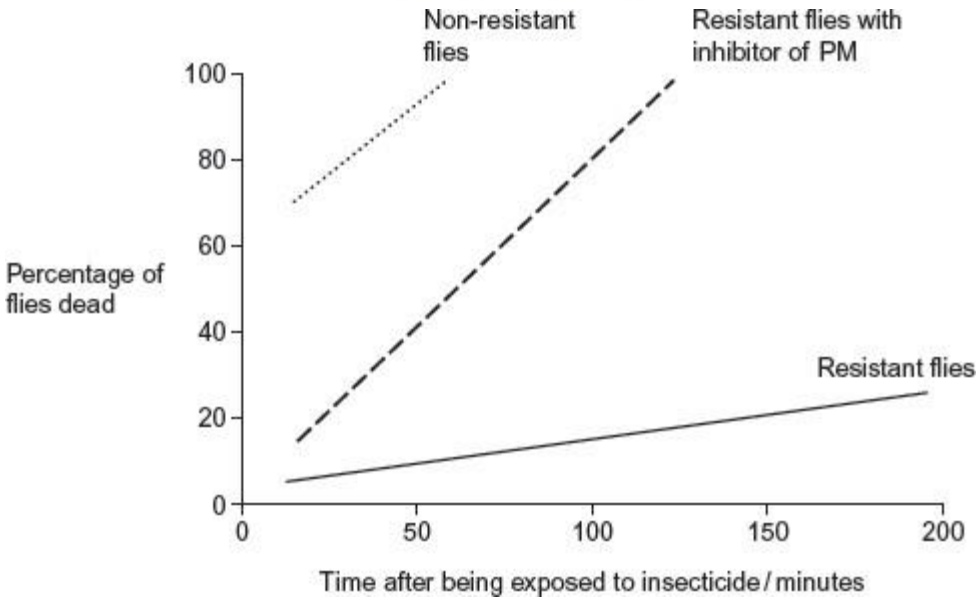
The scientists obtained large numbers of resistant and non-resistant flies. They then set up the following experiments.

- 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-resistantflies.

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(2)

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

Explain how these data support this conclusion.

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[Extra space] _____

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(4)

(Total 15 marks)

3 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**.

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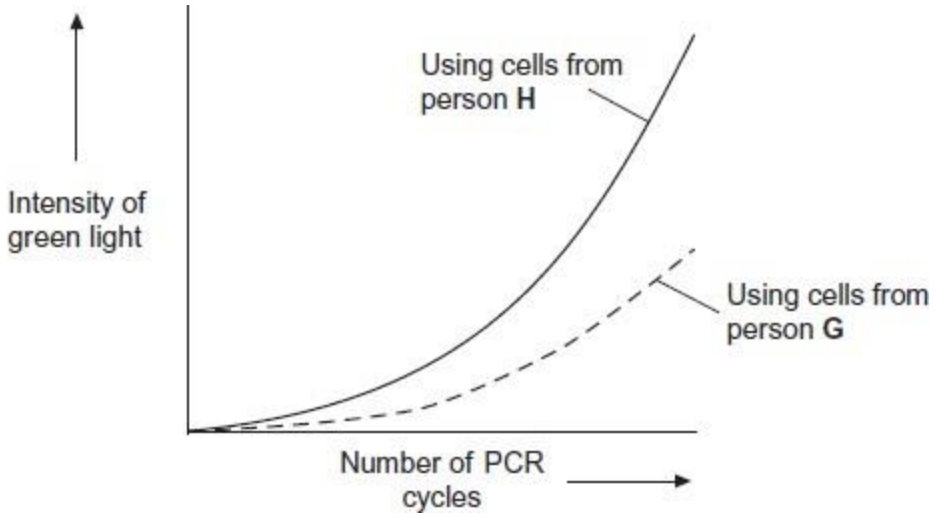
(2)

(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**.

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_____

(Extra space) _____

_____

_____

(3)

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

_____

_____

_____

_____

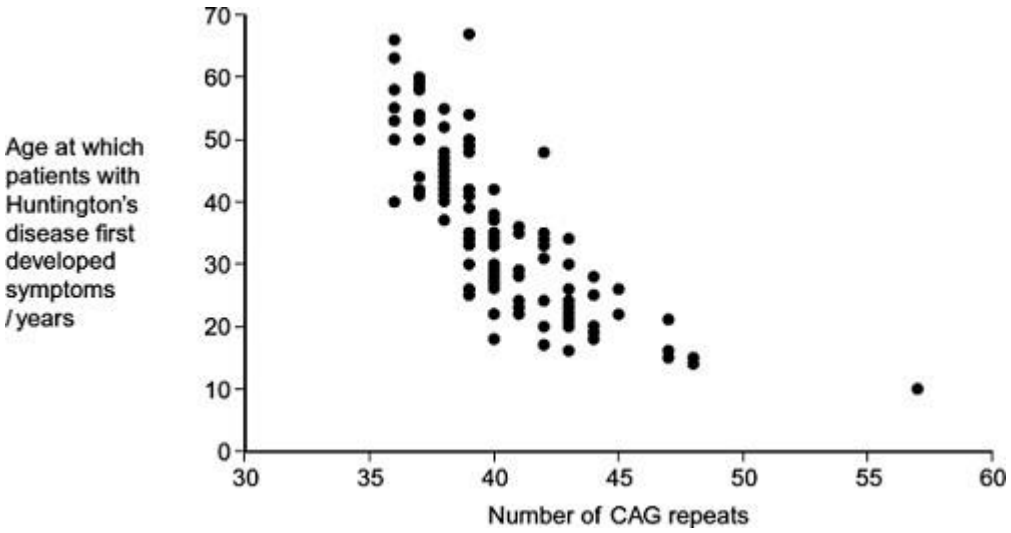
(2)

(Total 8 marks)

4 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.

_____

*(Extra space)*_____

_____

_____

(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.

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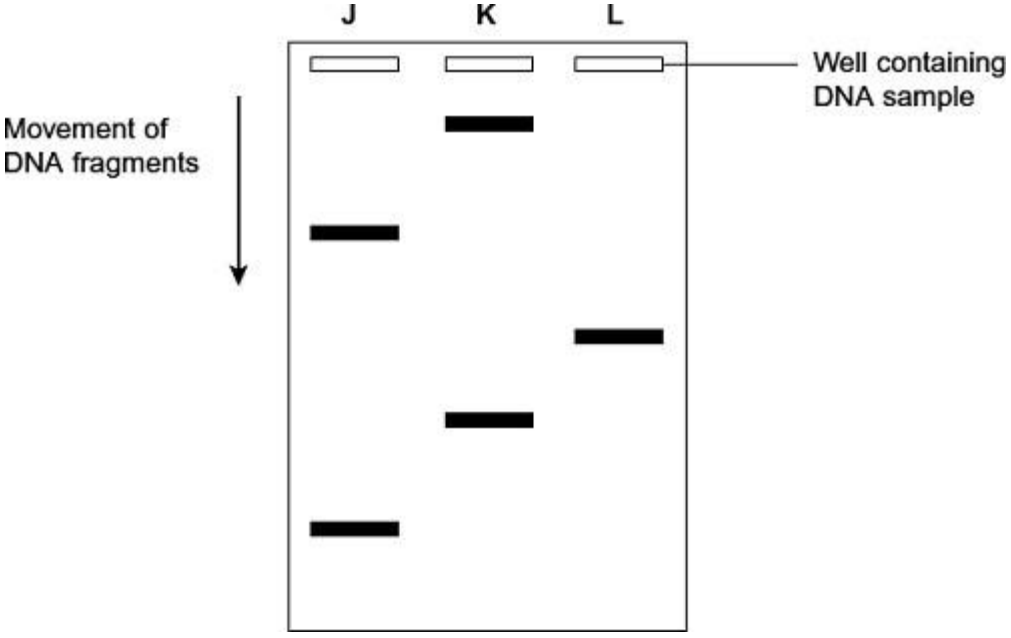
(2)

(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 wasthis? Explain your answer.

Person _____

Explanation _____

_____

_____

_____

(2)

(ii) The diagram only shows part of the gel. Suggest how the scientists found the numberof 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 wasseen for Person **L**.

_____

(1)

(Total 9 marks)

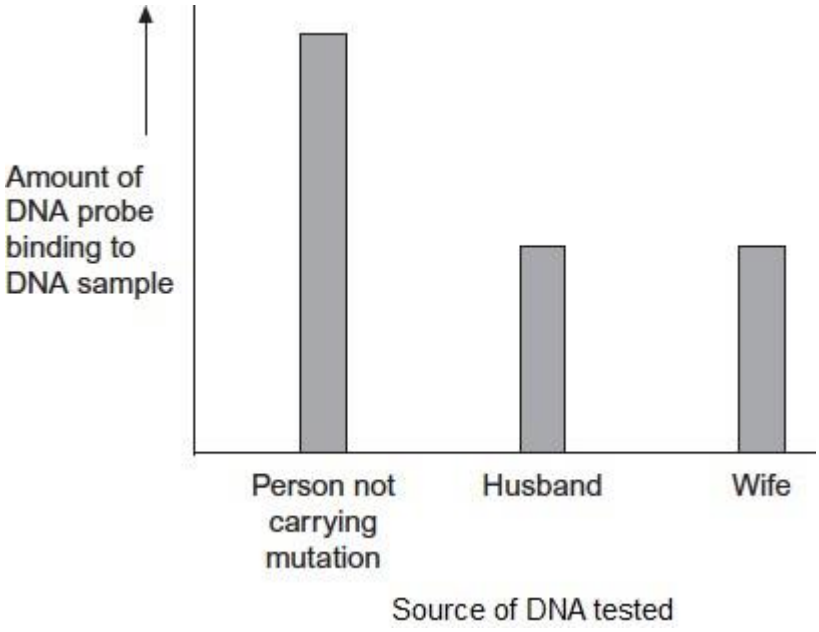
5 A husband and wife wanted to know whether they were carriers of the mutated form of a gene.

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.

*(Extra space)*_____

(3)

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

*(Extra space)*_____

(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)

(a) Scientists can use protein structure to investigate the evolutionary relationships between

6 different species. Explain why.

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(2)

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

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_____

(2)

(Total 4 marks)

Essay

7

You should write your essay in continuous prose.

Your essay will be marked for its scientific accuracy.

It will also be marked for your selection of relevant material from different parts of the specification and for the quality of your written communication.

The maximum number of marks that can be awarded is

Scientific	16
Breadth of knowledge	3
Relevance	3
Quality of written communication	3

Write an essay on the following topic:

Using DNA in science and technology

(Total 25 marks)

8 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

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_____

(1)

(ii) electrophoresis?

_____

_____

(1)

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

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(2)

(c) The scientists analysed the DNA on the Y chromosome and the DNA in the mitochondria ofthe 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)

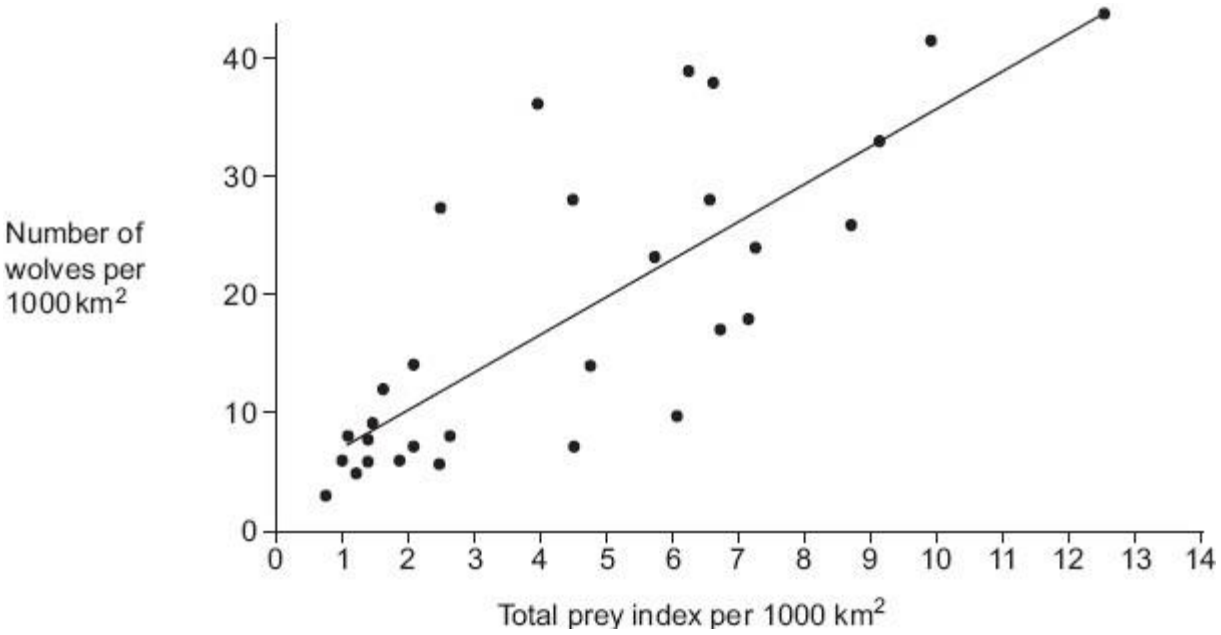
(ii) Suggest why DNA in the mitochondria helped them to reach this conclusion.

_____

_____

(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.

_____

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_____

(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.

_____

_____

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_____

(2)

(e) The ecologist calculated the total prey index by combining the prey indices and the totalnumber of animals of each species present in 1000 km^2. 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.

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(2)

(Total 12 marks)

9 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.

Stage 1 DNA is cut into fragments

↓

Stage 2 Electrophoresis separates the DNA fragments

↓

Stage 3 Radioactive DNA probes are used to locate specific DNA fragments

(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.

_____

_____

_____

_____

(2)

(Total 7 marks)

10 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

↓

Fragments separated from each other by electrophoresis

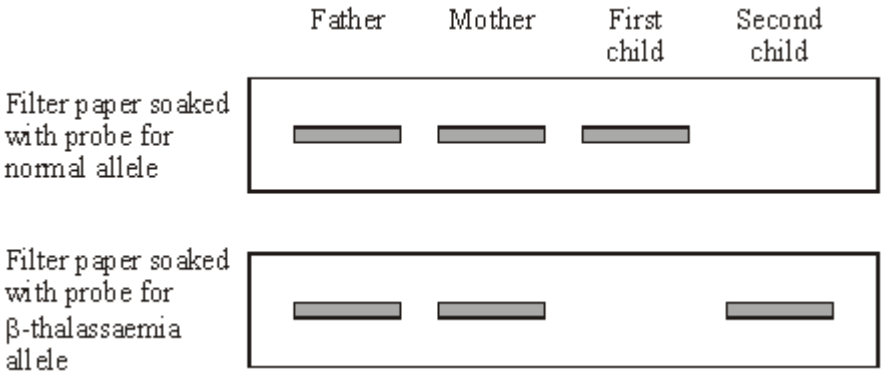
↓

One region of the resulting gel was blotted with two pieces of filter paper. The first was soaked in a solution containing a radioactive DNA probe for the normal allele. The second was soaked in a solution containing a radioactive DNA probe for the β-thalassaemia allele.

↓

Surplus probe washed off

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 hasβ-thalassaemia? Explain your answer.

(3)

(b) (i) The fragment of DNA containing the normal allele and the fragment with theβ-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.

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(2)

(Total 7 marks)

11 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**?

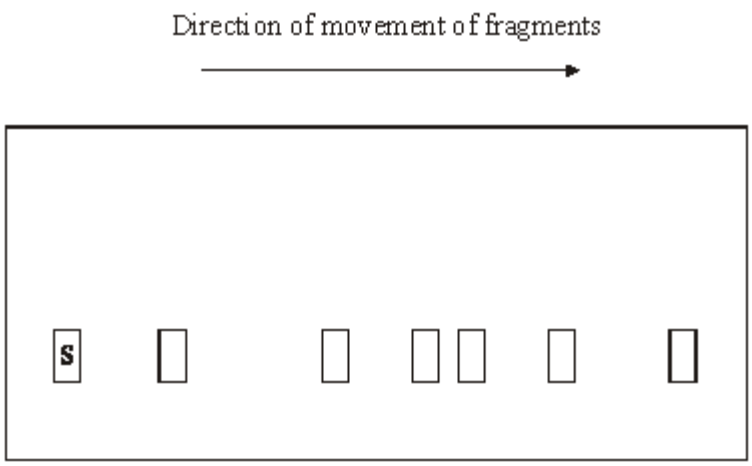
_____

(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.



(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)

Read the following passage.

12

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).

_____

_____

_____

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(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.

_____

_____

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(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 electrophoresisgel, 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)

13 Read the following passage.

Soon a single drop of blood might be enough to reveal, at a very early stage, if a patient has 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 5 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 ismore 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 ispresent.

_____

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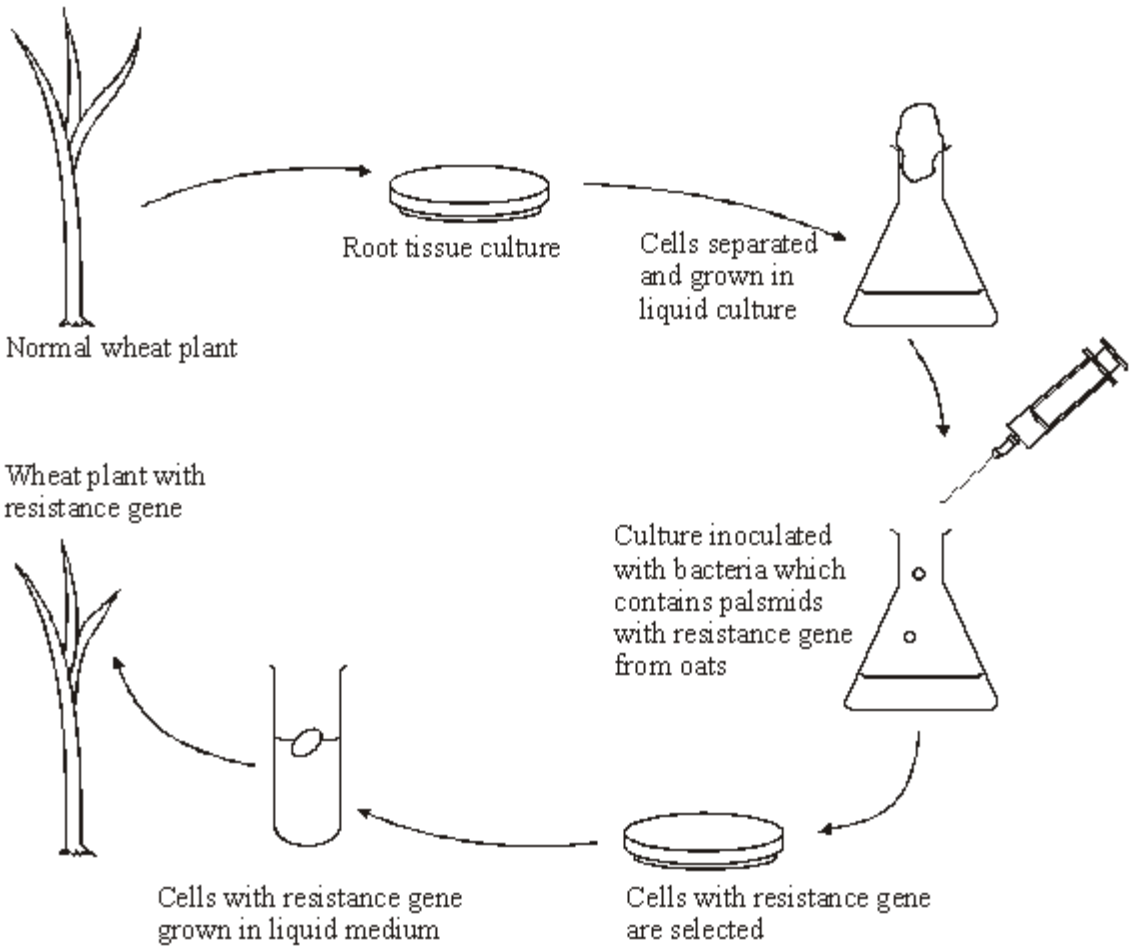
(3)

(Total 15 marks)

14 ‘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.

_____

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(2)

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

Describe how bacteria may be produced which have the resistance gene in their plasmids.

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(6)

(Total 10 marks)

15 (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.

Sample of DNA extracted from a person's tissue and heated to separate the strands

↓

Radioactive gene probe added to the DNA

↓

Excess probe washed away

↓

Sample tested for radioactivity

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

_____

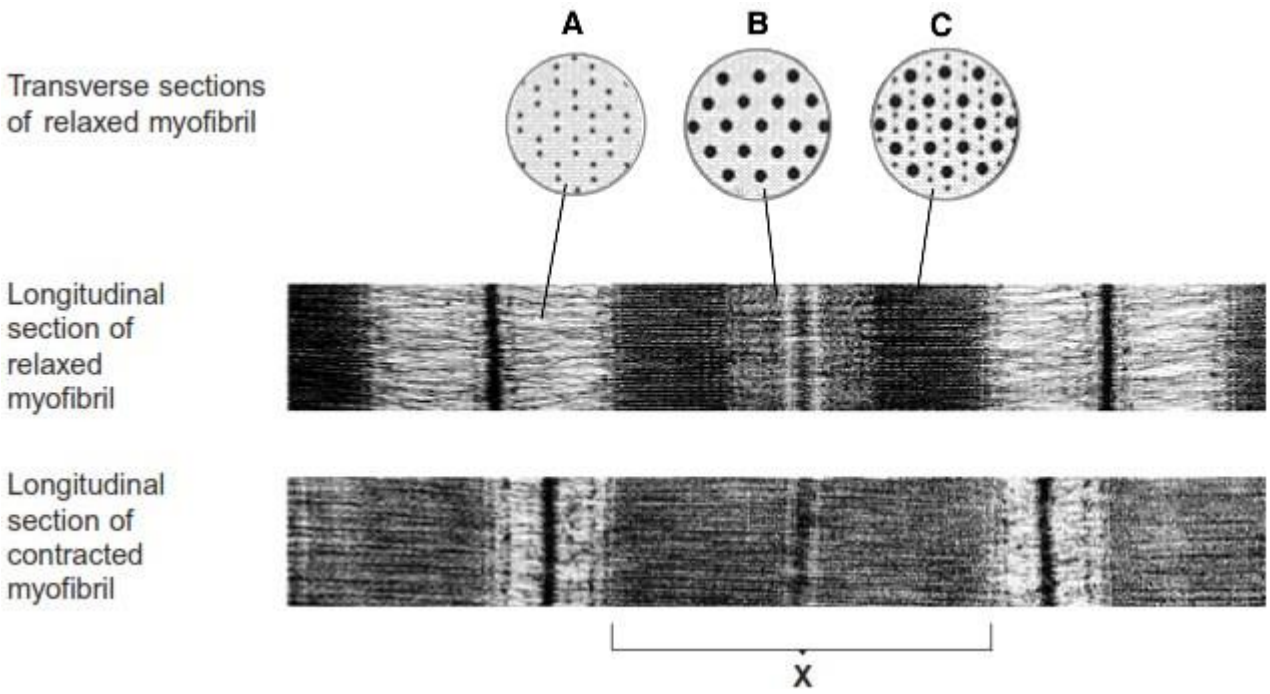
_____

(2)

(Total 6 marks)

16 **Figure 1** shows sections through relaxed and contracted myofibrils of a skeletal muscle. The transverse sections are diagrams. The longitudinal sections are electron micrographs.

Figure 1



(a) (i) The electron micrographs are magnified 40 000 times.
Calculate the length of band **X** in micrometres.
Show your working.

Length of band **X** = _____ µm

(2)

(ii) Explain the difference in appearance between transverse sections **A** and **C** in **Figure 1**.

_____

_____

(1)

(b) Explain what leads to the differences in appearance between the relaxed myofibril and the contracted myofibril.

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(*Extra space*) _____

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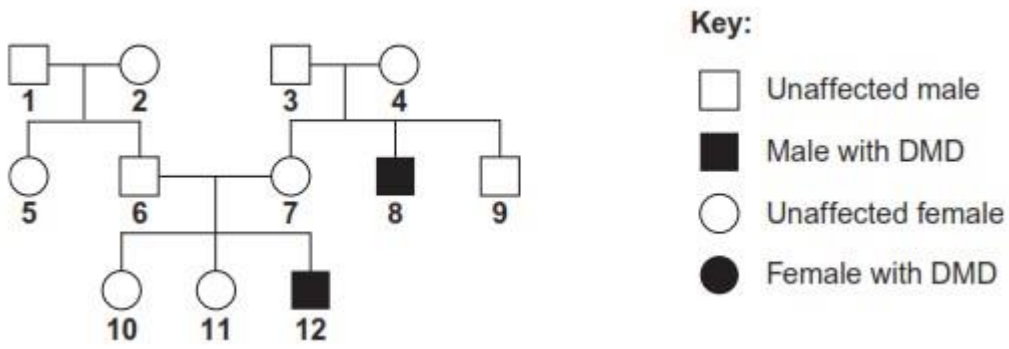
(4)

(c) Duchenne muscular dystrophy (DMD) is a condition caused by the recessive allele of a sex-linked gene. A couple have a son with DMD. They want to know the probability that they could produce another child with DMD. They consulted a genetic counsellor who produced a diagram showing the inheritance of DMD in this family. This is shown in **Figure 2**.

Figure 2



The couple who sought genetic counselling are persons **6** and **7**.

(i) Give the evidence to show that DMD is caused by a recessive allele.

_____

_____

(1)

(ii) Give the numbers of **two** people in **Figure 2** who are definitely carriers of muscular dystrophy.

_____

(1)

(iii) Complete the genetic diagram to find the probability that the next child of couple **6** and **7** will be a son with muscular dystrophy. Use the following symbols:

$\mathbf{X}^{D}$ = normal X chromosome

$\mathbf{X}^{d}$ = X chromosome carrying the allele for muscular dystrophy

Y = normal Y chromosome

	6	**7**
Parental phenotypes	Unaffected	Unaffected
Parental genotypes	_____	_____
Gametes	_____	_____

Offspring genotypes _____

Offspring phenotypes _____

Probability of having a son with DMD _____

(4)

(d) DMD is caused by a deletion mutation in the gene for a muscle protein called dystrophin. A deletion is where part of the DNA sequence of a gene is lost. People in different families may inherit mutations in different regions of this gene.

Scientists isolated the dystrophin gene from DNA samples taken from children **10**, **11** and **12**. They cut the gene into fragments using an enzyme. The scientists then used two DNA probes to identify the presence or absence of two of these fragments, called **F** and **G**. This allowed them to find the number of copies of each fragment in the DNA of a single cell from each child.

The table shows their results.

Child	**Number of copies of gene fragment per cell**	
	F	**G**
10 (unaffected girl)	2	1
11 (unaffected girl)	2	2
12 (boy with DMD)	1	0

(i) The number of copies of gene fragments **F** and **G** shows that person **12** has DMD. Explain how.

_____

_____

(1)

(ii) The number of copies of gene fragments **F** and **G** shows that person **12** is male. Explain how.

_____

_____

(2)

(iii) The genetic counsellor examined the scientists' results. He concluded that person **10** is a carrier of DMD but her sister, **11**, is not.

Describe and explain the evidence for this in the table.

(*Extra space*)

(3)

(e) Person **12** took part in a trial of a new technique to help people with DMD.

Doctors took muscle cells from person **12**'s father and grew them in tissue culture.

They suspended samples of the cultured cells in salt solution and injected them into a muscle in person **12**'s left leg. They injected an equal volume of salt solution into the corresponding muscle in his right leg. Person **12** was given drugs to suppress his immune system throughout the trial.

Four weeks later, the doctors removed a muscle sample from near the injection site in each leg. They treated these samples with fluorescent antibodies. These antibodies were specific for the polypeptide coded for by gene fragment **G** of the dystrophin gene.

The results are shown in the table.

Location and treatment	Percentage of muscle fibres labelled with antibody
Left leg - injected with cultured cells suspended in salt solution	6.8
Right leg - injected with salt solution	0.0

(i) Why was it necessary to treat person **12** with drugs to suppress his immune system?

_____

_____

(1)

(ii) Explain why salt solution was injected into one leg and cultured cells suspended insalt solution into the other.

_____

_____

_____

(1)

(iii) This technique is at an early stage in its development. The doctors suggested thatfurther investigations need to be carried out to assess its usefulness for treating people with DMD.

Explain why they made this suggestion.

_____

(*Extra space*) _____

(4)

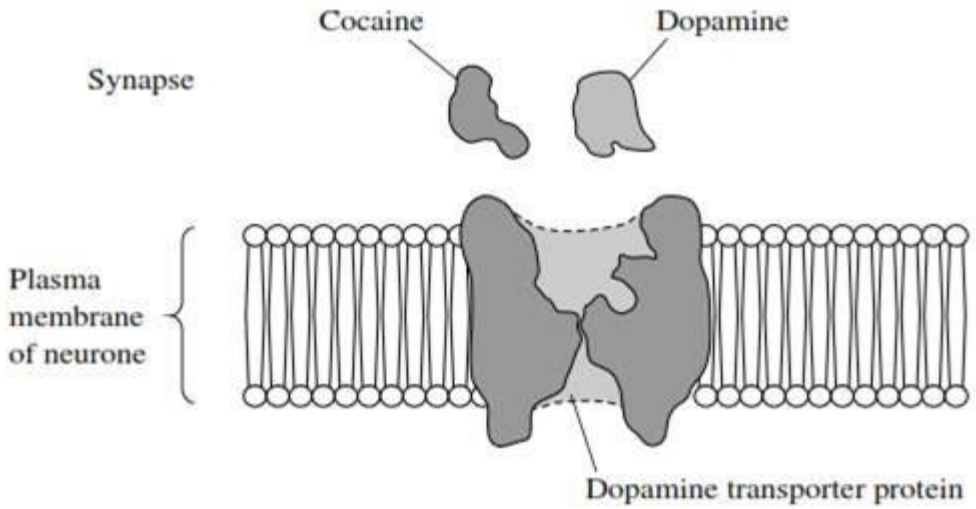
(Total 25 marks)

17 Cocaine is a highly addictive and illegal drug.

The release of the neurotransmitter dopamine in specific synapses in the brain leads to feelings of pleasure. Dopamine is removed from synapses by dopamine transporter proteins in the plasma membrane of neurones. Cocaine binds to the dopamine transporter protein.

Figure 1 shows a dopamine transporter protein and molecules of cocaine and dopamine.

Figure 1



(a) Using all of the information, suggest how cocaine leads to feelings of pleasure.

_____

_____

_____

_____

_____

_____

(Extra space) _____

_____

_____

(3)

(b) (i) Scientists isolated a mutated gene for the dopamine transporter protein.

Name **one** method that the scientists could have used to produce many copies of the mutated gene in the laboratory.

_____

(1)

(ii) Copies of the gene were then inserted into early embryos of mice. When these mice were born, samples of their DNA were tested using DNA probes to make sure that the mutated gene was present in the mice.

What is a DNA probe?

_____

_____

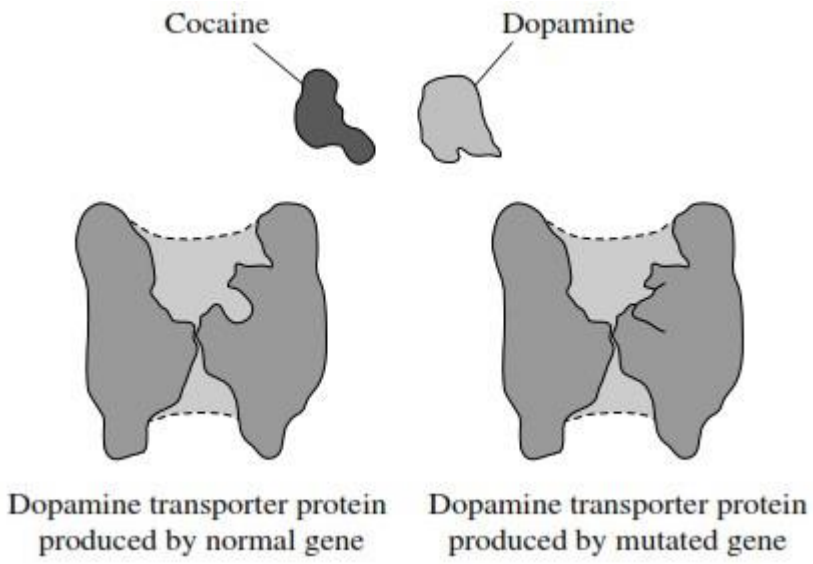
_____

_____

(2)

(c) **Figure 2** shows dopamine transporter proteins produced from the normal gene and from the mutated gene.

Figure 2



Explain how the mutation leads to the production of a protein that transports dopamine but is **not** affected by cocaine.

_____

_____

_____

_____

_____

_____

(Extra space) _____

_____

_____

(3)

(Total 9 marks)

Mark schemes

1

(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) andglows/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/ seewhich 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/ totrace 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]

2

(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 pairfragments;

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 pairfragments.

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 thannon-resistant flies.

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

4 max

[15] (a) Reverse transcriptase;

3 **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 meansmore light;

2. DNA (with **A**) doubles each (PCR) cycle;

3. So light (approximately) doubles / curve steepens more and more (eachcycle) / 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] (a) (i) 1. Negative correlation;

4

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

1

(iii) Homozygous / (CAG) fragments are the same length / size / mass;

Accept: small fragment has run off gel / travelled further

1

[9

] (a) 1. Carriers are heterozygous / have one normal copy and one mutant copy of gene /

5 have one recessive allele / don't have the condition;

2. Both have DNA that binds (about) half / 50% amount of probe (that non-carrierdoes);

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 donot 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) faultyprotein / 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] (a) 1. Closer the (amino acid) sequence the closer the relationship;

6

2. (Protein structure) related to (DNA) base / triplet sequence;

Amino acid sequence is related to (DNA) base / triplet sequence = two marks;

2

(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

[4]

Essay Using DNA in science and technology

7

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.

(a) (i) To cut the DNA;

8

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 / malegamete;

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]

9

(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]

10

(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]

11 (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]

12 (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] (a) 1 (DNA altered by) mutation;

13

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;

2

(c) normal DNA and changed DNA have different sequences;
DNA only binds to complementary sequence;

2

(d) fewer abnormal / cancerous cells / smaller tumours;less cell damage / less spread / fewer locations to treat;

2

(e) mRNA base sequence has changed;gene / DNA structure is different / has mutated; cancer gene active / tumour suppressor gene inactive;

3

[15] (a) (i) contains genes / nucleotides / sections of DNA / artificial

14

DNA from two species / 2 types of organisms;

1

(ii) carries gene / DNA (into the other organism / gene carrier);

1

(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;

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] (a) probe will attach (to mutant allele);

15

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] (a) Correct answer: 1.25;

16

Ignore working

OR (if wrong answer)

$$\frac{\text{measurement in µm}}{40000} \; / \; \frac{\text{measurement in mm}}{40} = 1 \text{ mark}$$

125 but wrong order of magnitude = 1 mark

2 (ii) **C** has myosin / thick (and actin / thin) filaments;

OR

A has only actin / thin (/ no myosin / no thick) filaments;

1 max

(b) When contracted:

Thick & thin filaments/myosin & actin overlap more;

Interaction between myosin heads & actin / cross-links form;

Movement of myosin head;

Thin filaments / actin moved along thick filaments / myosin;

Movement of thin filaments / actin pulls Z-lines closer together;

Displacement of tropomyosin to allow interaction;

Role of Ca^{2+};

Role of ATP;

Allow ref. to 'sliding filament mechanism' / described if no other marks awarded

4 max

(c) (i) 8 has DMD but 3 and 4 do not / 12 has DMD but 6 and 7 do not / neither parent has the condition but their child has;

Allow parents 3 and 4 give 8, parents 6 and 7 give 12

1

(ii) 4 ***AND*** 7;

1 (iii) Parental genotypes: 6 = $\mathbf{X^DY}$ AND 7 = $\mathbf{X^DX^d}$

AND

Gametes correct for candidate's P genotypes – e.g.

$\mathbf{X^D}$ and **Y** + $\mathbf{X^D}$ and $\mathbf{X^d}$;

Offspring genotypes <u>correctly derived</u> from gametes e.g.

$\mathbf{X^DX^D}$ + $\mathbf{X^DX^d}$ + $\mathbf{X^DY}$ + $\mathbf{X^dY}$;

Male offspring with MD correctly identified: $\mathbf{X^dY}$;

Probability = 0.25 / correct for candidates offsprings genotypes;

Accept ¼ / 1 in 4 / 1:3 / 25%

NOT '3:1' / '1:4'

4

(d) (i) No gene fragment **G**;

1

(ii) Only one copy of gene fragment **F**;

Male has only one X-chromosome / is XY
(c.f. female has two / is XX);

2

(iii) 10 has only one copy of gene fragment **G**;

10 has only one normal X-chromosome / has one abnormal / X^{dDd} has only one normal allele / has one X / is X X / is heterozygous;

11 has two normal X-chromosomes / has 2 normal alleles / is $X^{DD}X$ / has not got X^{d} / has 2 copies of (F and) G;

3

(e) (i) To prevent rejection / prevent antibody production vs. injected cells / injected cells have (foreign) antigen (on surface);

1

(ii) Shows effect of <u>cells</u> / not just effect of injection / not just effect of salt solution;

1

(iii) Only one person tested so far – need more to see if similar results /need more to see if reliable;

Need to assess if new (dystrophin positive) muscle fibres are functional / if muscle becomes functional;

Can't tell how widespread effect is in the muscle / sample taken near injection site;

Need to test for harmful side effects;

Need to test if successful for other mutations of dystrophin gene;

Need to assess permanence / longevity of result/insufficient time allowed in investigation;

(In this patient) only small response / %;

Further sensible suggestion;

4 max

[25] (a) Cocaine (binding) changes shape of transporter/prevents dopamine binding;

17

Reject references to active site

Transporter cannot move (bound) dopamine (through membrane / protein / into cell);
Dopamine remains / builds up in synapses (leading to feelings of pleasure);

3

(b) (i) Polymerase chain reaction / PCR;

1

(ii) Single-stranded DNA;

Reject reference to a single strand of DNA

Bases / sequence complementary to DNA / gene to be identified;

(Radioactively / fluorescent) labelled so that it can be detected;

2 max

(c) Mutation changes base sequence of gene / DNA;

Accept references to active site

(Thus) changing amino acid sequence;
Changes tertiary structure / shape of protein/transporter;

Cocaine binding site changes/cocaine cannot bind;
Dopamine can still bind (and be transported);

3 max

[9]