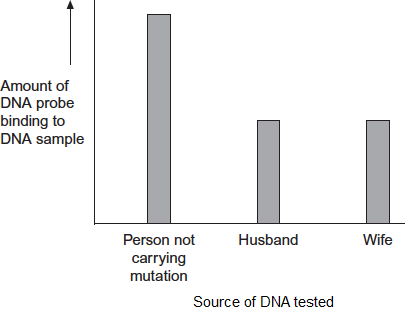
**Q1.**A husband and wife wanted to know whether they were carriers of the mutated form of a gene. This mutation is a deletion that causes a serious inherited genetic disorder in people who are homozygous.

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.

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**(3)**

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

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**(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?

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**(2)**

**(Total 8 marks)**

**Q2.**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.

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**(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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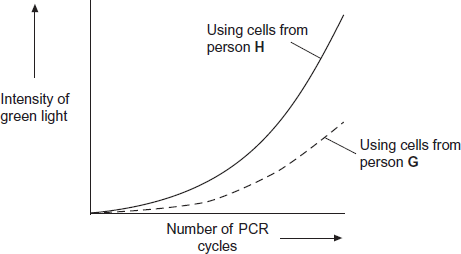
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**(2)**

(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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**(3)**

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

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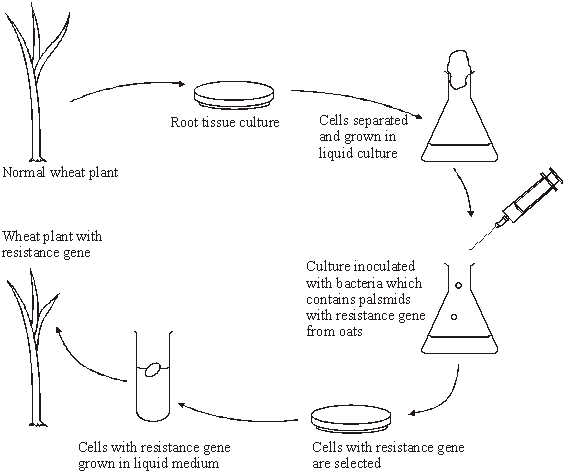
**(2)**

**(Total 8 marks)**

**Q3.** ‘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?

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**(1)**

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

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**(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)**

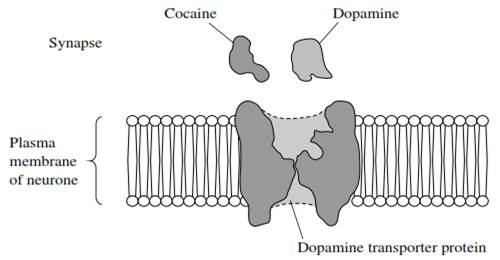
**(Total 10 marks)**

**Q4.**          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.

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*(Extra space) ................................................................................................*

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**(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.

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**(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?

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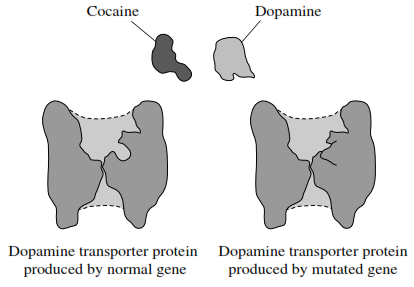
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**(2)**

(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.

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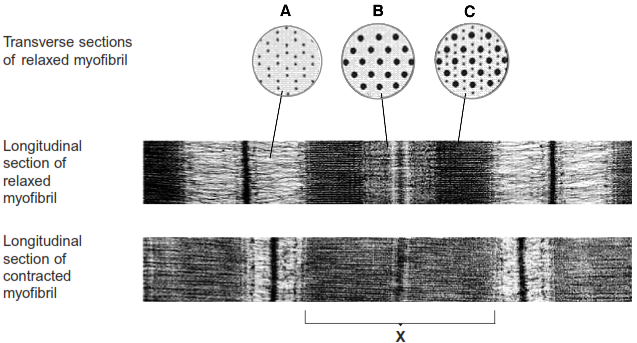
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**(3)**

**(Total 9 marks)**

**Q5.**          **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**.

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**(1)**

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

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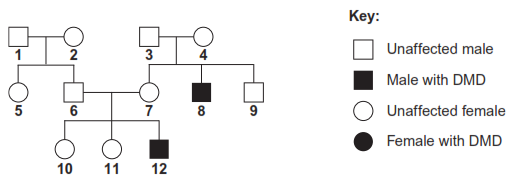
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**(4)**

(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.

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**(1)**

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

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**(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:

**X**D= normal X chromosome  
**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.

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**(1)**

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

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**(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.

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**(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?

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**(1)**

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

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**(1)**

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

Explain why they made this suggestion.

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**(4)**

**(Total 25 marks)**

**Q6.**          (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 |

**↓**

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| --- |
| 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.

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**(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

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Against

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**(2)**

**(Total 6 marks)**

**Q7.**          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.

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**(6)**

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

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**(2)**

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

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**(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.

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**(2)**

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

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**(3)**

**(Total 15 marks)**

**Q8.**          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**.

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**(1)**

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

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**(2)**

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

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**(2)**

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

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**(2)**

**(Total 7 marks)**

**Q9.**          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

5     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.

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

15   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.

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**(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.

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**(2)**

(c)     Describe the polymerase chain reaction.

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**(6)**

**(Total 15 marks)**

**Q10.**          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**?

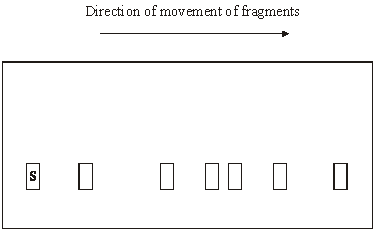
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**(1)**

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

|  |  |
| --- | --- |
| **Fragment** | **Number of base pairs (× 103)** |
| 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.

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**(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.

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**(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?

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**(1)**

**(Total 6 marks)**

**Q11.**          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 |

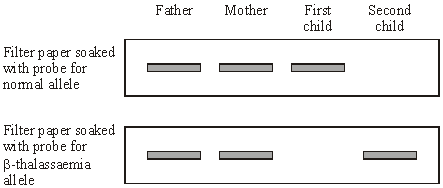
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|  |
| --- |
| One region of the resulting gel was blotted with two pieces of filter paper. The first was soaked in a solution containing a radioactive DNA probe for the normal allele. The second was soaked in a solution containing a radioactive DNA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

**↓**

|  |
| --- |
| Surplus probe washed off |

The diagram belowshows 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.

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**(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.

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**(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)**

**Q12.**          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?

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**(1)**

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

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**(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.

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**(1)**

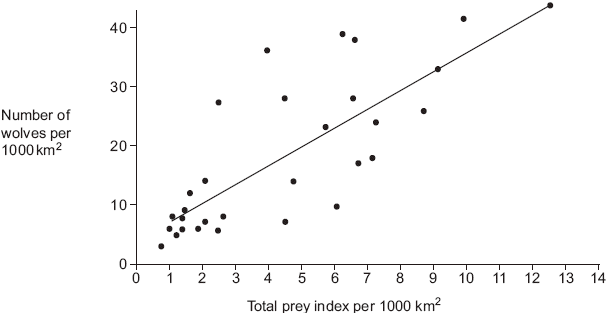
(ii)     Suggest why DNA in the mitochondria helped them to reach this conclusion.

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**(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 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.

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**(2)**

**(Total 12 marks)**

**Q13.Essay**

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

Write an essay on the following topic:

Using DNA in science and technology

**(Total 25 marks)**

**Q14.**(a)     Scientists can use protein structure to investigate the evolutionary relationships between 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)**

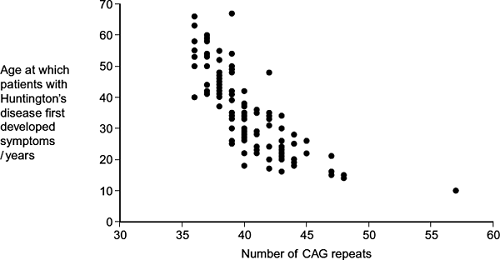
**Q15.**          Huntington’s disease is a genetic condition that leads to a loss in brain function. The gene involved contains a section of DNA with many repeats of the base sequence CAG. The number of these repeats determines whether or not an allele of this gene will cause Huntington’s disease.

•        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.

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**(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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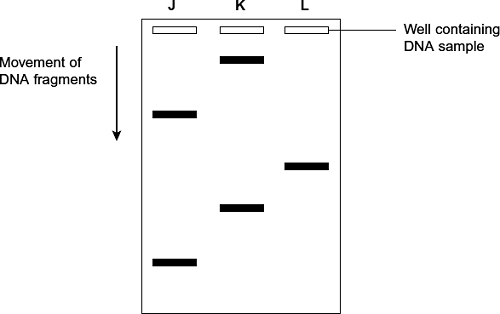
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**(2)**

(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 was this? Explain your answer.

Person ..................................................................................................

Explanation ...........................................................................................

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**(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.

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**(1)**

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

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**(1)**

**(Total 9 marks)**

**Q16.**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.

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**(2)**

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

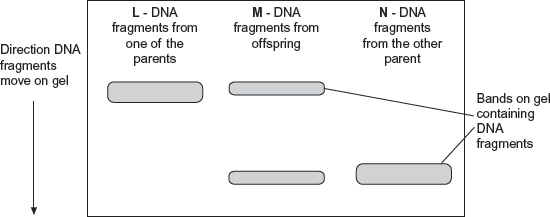
•        Primer A3 only binds to a 195 base-pair fragment from allele **r**.

•        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**

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(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]** ................................................................................................

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**(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.

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**(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.

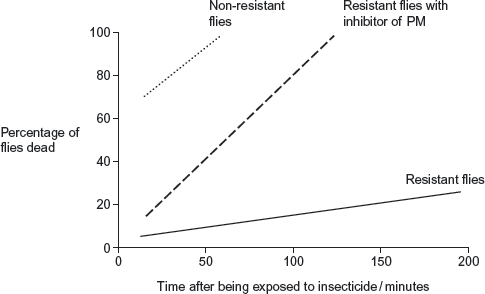
•        Resistant flies exposed to insecticide.

•        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**

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

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**(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.

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**(4)**

**(Total 15 marks)**

**M1.**(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]**

**M2.**(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]**

**M3.**          (a)     (i)      contains genes / nucleotides / sections of DNA / artificial  
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 Ca2+ / calcium salts / electric shock;  
                   (if ref. to ‘insulin’ allow 5 max.)

**max 6**

**[10]**

**M4.**         (a)     Cocaine (binding) changes shape of transporter/prevents dopamine binding;

*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]**

**M5.**          (a)     Correct answer: 1.25;

*Ignore working*

***OR*** (if wrong answer)

 / = 1 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 Ca2+;

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 = **XDY** AND 7 = **XDXd**

***AND***

Gametes correct for candidate’s P genotypes ‒ e.g.

**X**Dand **Y** + **X**Dand  **X**d**;**

Offspring genotypes correctly derived from gametes e.g.

**X**D**X**D+ **X**D**X**d+ **X**D**Y** + **X**d**Y**;

Male offspring with MD correctly identified: **X**d**Y**;

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 /  
has only one normal allele / has one Xd / is XDXd / is heterozygous;

11 has two normal X-chromosomes / has 2 normal alleles /  
is XDXD / has not got Xd / 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 cells / 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]**

**M6.**          (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]**

**M7.**          (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;

**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]**

**M8.**          (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]**

**M9.**          (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]**

**M10.**          (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]**

**M11.**          (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]**

**M12.**          (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]**

**M13.**          **Essay Using DNA in science and technology**

**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.

**M14.**(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;*

**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]**

**M15.**         (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*

**1**

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

*Accept: small fragment has run off gel / travelled further*

**1**

**[9]**

**M16.**(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**

**[15]**

**E1.**(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.

**E2.**(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.

**E3.**          (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.

**E4.**          This question was answered well by many candidates. It was pleasing to see that most were able to interpret the information presented in the diagrams. In (a), about a fifth of candidates obtained all three marks and three fifths obtained one or two marks. Weaker candidates appeared not to have read the stem of the question carefully and some seemed to think that the transporter protein was an enzyme that made dopamine. Others thought that cocaine entered the cell instead of dopamine and produced the same effect as dopamine inside the cell. Part (c) was particularly well answered and over a third of candidates obtained all three marks.

**E5.**          (a)     Most candidates measured band X (the A-band in an electron micrograph of a myofibril) correctly. Many did not then understand that they had to divide this by the stated magnification. Among those who did, many had problems interconverting millimetres and micrometres and were often several orders of magnitude out. Only one quarter of candidates were entirely successful.

In part (ii), most candidates knew the correct distribution of actin and myosin filaments in the two distinct bands of the myofibril. One unusual, and erroneous, concept expressed by a number of candidates was that one part of the myofibril was contracted at the same time as the other part was relaxed.

(b)     Many candidates gave a full and clear account of the process of muscle contraction, including the roles of ATP, calcium ions, tropomyosin, the attachment of the myosin head to actin and its movement causing the actin filament to slide along the myosin. Weaker candidates just described how the appearance of the various bands changed when the myofibril contracted rather than offering the required explanation. Almost one-third of candidates scored full marks.

(c)     Using information from the pedigree diagram showing the inheritance of Duchenne muscular dystrophy (DMD) over three generations, almost two-thirds of candidates cited the production of a child with muscular dystrophy by unaffected parents as evidence for the condition being caused by a recessive allele. However, less than half the candidates were able to identify two carriers from the diagram.

In completion of the genetic diagram, common errors included switching the genders of the two parents, giving the male parent a genotype that would have resulted in him having muscular dystrophy, incomplete assignment of phenotypes to the offspring genotypes (both gender and having / not having DMD were important) and, having shown that 25% of the offspring would be expected to be male with DMD, to then halve this figure to 12.5 %. Additional, incorrect, answers on the probability line, e.g., ‘25% or 1 : 4’, failed to gain the mark. Despite this, almost one-third of candidates scored full marks in this section.

(d)     Just over half the candidates answered part (i) correctly, realising that the complete absence of one of the gene fragments indicated that the person would suffer from DMD. In part (ii), these candidates realised it was the single copy of the other gene fragment (compared with two copies in each of his sisters) that indicated the person concerned was male as he had just one X-chromosome while his sisters had two. Only about one-fifth of candidates were able to tell the complete story, although some two-thirds got half-way.

Part (iii) differentiated very well between candidates who gave varying degrees of appropriate detail in their answers. The most able noticed that one of the girls had two copies of one of the gene fragments while her sister, having but a single copy of this fragment, must have been the carrier as she would have had one normal X chromosome (hence being healthy herself) and one carrying the mutation responsible for DMD. Approximately one quarter scored full marks, although nearly two-thirds were able to make at least two of the three points required.

(e)     Far too many candidates failed to use appropriate terminology in part (i). There were no marks available for stating that the ‘immune system’ (given in the question) ‘fought against’ / ‘attacked’ the implanted cells. Terms such as *rejection*, *antibody* and *antigen* were required. Less than half the candidates used such terms.

Similarly, in part (ii), there was no mark available for merely stating that the injection with salt solution served as a ‘control’. The purpose of the control was required, e.g., so that the effect of the cells injected into the other leg became apparent, or to show it was not just the salt solution that had caused the effect in the other leg. Approximately half the candidates gave the appropriate detail.

In part (iii), there was plenty of scope for candidates to explain the limitations of the given investigation and to suggest appropriate further work that could be done. Candidates made general points about the limited sample size (i.e., just *one* individual), the short time period allowed to assess the effect of the treatment, or they made specific points relating to the given size of the response, the fact that success had so far been achieved only for this particular mutation, that only a measure of the *presence* of the appropriate type of muscle cells had been performed with no information about their ability to function, etc. The question differentiated very well amongst candidates who took varying amounts of care in selecting information, in assessing the reliability of the data and in applying their knowledge and understanding of how an investigation should be carried out in order to obtain reliable results and to draw valid conclusions. Although almost 90% of candidates were able to make at least one valid point, only 3% scored all 4 marks.

**E6.**          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.

**E7.**          (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.

**E8.**          (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.

**E9.**          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.

**E10.**          **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.

**E11.**          (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.

**E12.**          (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.

**E13.**          **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.

**E14.**(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.

**E15.**         (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.

**E16.**(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.