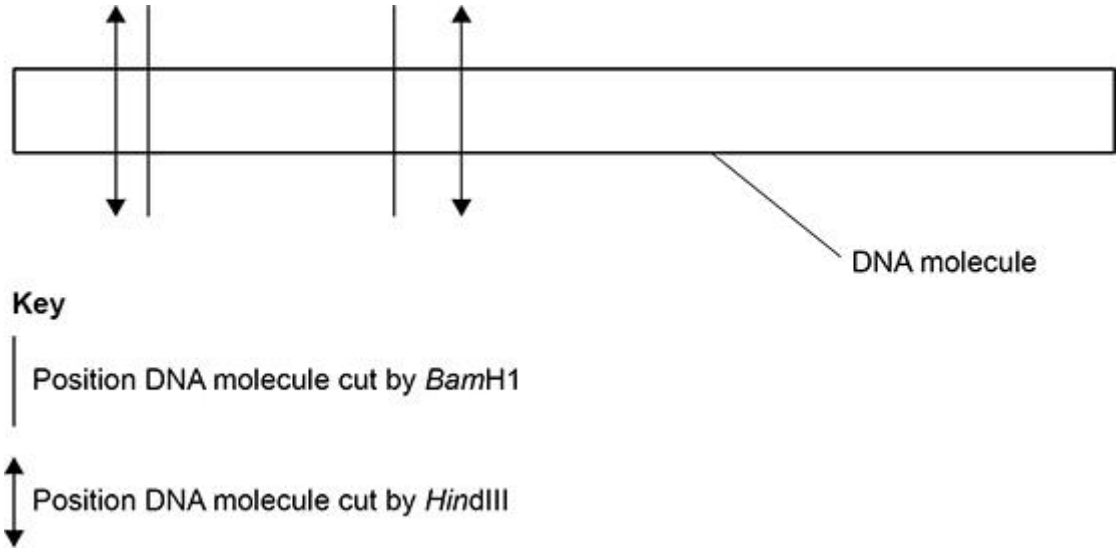
## Q1.

(a) *Bam*H1 and *Hin*dIII are both restriction endonucleases.

**Figure 1** shows the positions where these enzymes cut a linear molecule of DNA.

**Figure 1**



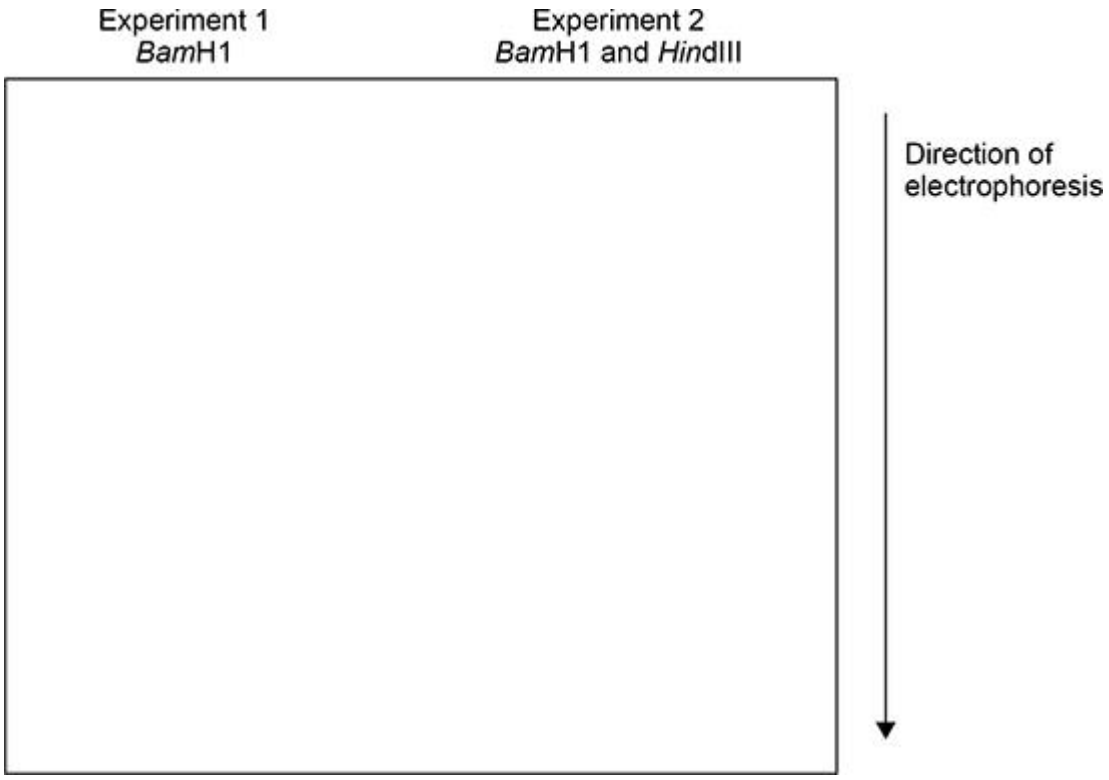

In two experiments, multiple copies of the DNA molecule shown in **Figure 1** were all completely cut into fragments using these restriction enzymes. The DNA fragments produced were then separated by electrophoresis.

Experiment 1 – DNA cut into fragments using *Bam*H1.

Experiment 2 – DNA cut into fragments using *Bam*H1 and *Hin*dIII.

Complete **Figure 2** to show the relative positions of the bands following electrophoresis in experiments 1 and 2.

**Figure 2**




**(2)**

(b) Suggest how you could determine the size of the different DNA fragments produced in these experiments.

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**(2)**

(c) How many DNA fragments would be produced in experiment 2 if the original DNA molecule was a plasmid?

_____

**(1)**

(d) Describe how restriction endonuclease and DNA ligase are used to insert a gene into a plasmid.

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**(2)**

**(Total 7 marks)**

## Q2.

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

STRs are found throughout the whole genome.

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

(a) Describe how STRs could be removed from a sample of DNA.

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**(2)**

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

Explain the importance of knowing these base sequences in PCR.

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**(2)**

(c) A single STR molecule consisting of a 12 **base pair** sequence of AGAT was amplified 50 times using PCR.

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

Show your working.

Answer _____

**(2)**

(d) During genetic fingerprinting, the different STRs are separated by gel electrophoresis.

Give **two** features of STRs which enable them to be separated by gel electrophoresis.

1 _____

_____

2 _____

_____

**(2)**

**(Total 8 marks)**

## Q3.

Read the following passage.

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

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

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

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

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

(a) *BRCA1* and *BRCA2* are human genes that code for tumour suppressor proteins. Mutations in *BRCA1* and *BRCA2* can cause cancer (lines 1–2). Explain how.

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**(3)**

(b) Genetic testing, using DNA from saliva, can screen for all known harmful mutations in both genes (lines 5–6). Describe how this DNA could be screened for all known harmful mutations in both genes.

**(4)**

(c) Effective treatment of ER-positive breast cancers often involves the use of drugs which have a similar structure to oestrogen (lines 9–10).

Suggest and explain how these drugs are an effective treatment of ER-positive breast cancers.

**(3)**

(d) Blood tests can be used to test for cancers (line 11). However, the results of blood tests may **not** be conclusive when testing for prostate cancer. Explain why.

**(2)**

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

**(3)**

**(Total 15 marks)**

## Q4.

(a) Complete the following definitions.

The genome is _____

_____

_____

The proteome is _____

_____

_____

**(2)**

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

(b) Give **two** reasons why bacteria are able to use human DNA to produce human proteins.

1 _____

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2 _____

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**(2)**

(c) Suggest and explain **one** reason why bacteria might **not** be able to produce every human protein.

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**(1)**

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

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

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

The diagram below shows an example of a DNA fragment that can be transferred into the cells of goats.


(d) The enhancer stimulates region **M**.

Name regions **M** and **N** shown in above diagram.

Region **M** _____

Region **N** _____

**(2)**

(e) Explain the purpose of the marker gene.

_____

**(1)**

(f) The enhancer only stimulates region **M** in the milk-producing glands of a goat.

Suggest **two** explanations for the importance of the enhancer being included in the DNA fragment transferred.

1 _____

_____

_____

_____

2 _____

_____

_____

_____

**(2)**

**(Total 10 marks)**