DNA Probes and Genetic Fingerprinting: AQA A Level Biology Exam Que

12 questions · 28 marks · about 36 minutes. Answer in the spaces, then check the mark schemes at the end.

Question 1 · A level Paper 2 · June 2019 · Q08.1 · 2 marks

0 8 . 1 What is a DNA probe?

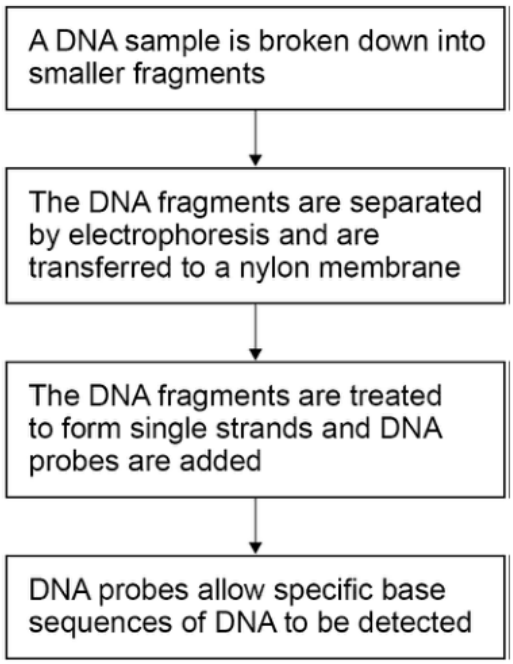
[2 marks]

Question 2 · information from earlier in the question

DNA probes are used to detect specific base sequences of DNA.

The process is shown in **Figure 6**.

Figure 6



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Question 2 · A level Paper 2 · June 2019 · Q08.3 · 1 mark

0 8 . 3 The DNA on the nylon membrane is treated to form single strands. Explain why.

[1 mark]

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Question 3 · A level Paper 3 · November 2021 · Q06.1 · 4 marks

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

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

0 6 . 1 The scientists used a radioactively labelled DNA probe to show that the cells of tobacco plant leaves contained the *SUT1* gene.

Describe how they would do this.

Do **not** include PCR in your answer.

[4 marks]

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Question 4 · information from earlier in the question

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

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

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

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

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

Question 4 · A level Paper 2 · June 2017 · Q10.7 · 2 marks

1 0 . 7 Suggest why a DNA probe for the mutated triplet was **not** considered a suitable method for detection of the E280A mutation (lines 22–23).

[2 marks]

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Question 5 · information from earlier in the question

1 0 Read the following passage.

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

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

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

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

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

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Question 5 · A level Paper 2 · June 2022 · Q10.2 · 4 marks

1 0 . 2 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 marks]

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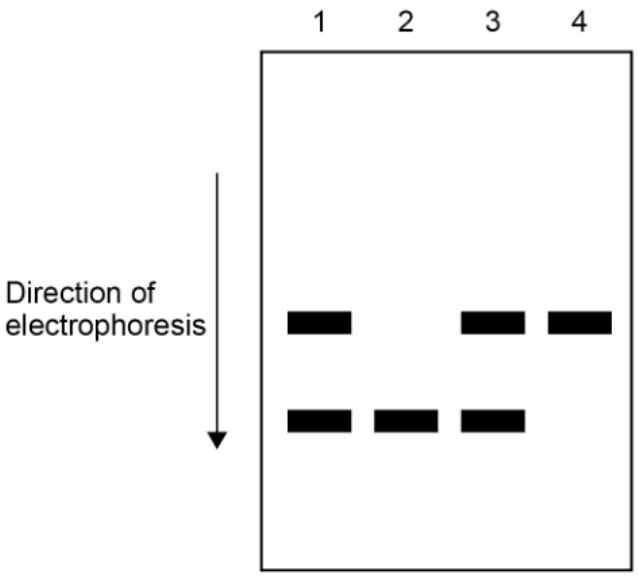
Question 6 · information from earlier in the question

CF can be caused by a three-base deletion mutation in the *CFTR* gene. This deletion creates a recessive allele called F508del.

DNA probes and gel electrophoresis were used to screen two parents and their two children for the F508del allele. One of the children was diagnosed with CF.

Figure 5 shows the results.

Figure 5



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Question 6 · A level Paper 2 · June 2025 · Q09.3 · 3 marks

0 9 . 3 The identity of each family member can be determined from the relative position of the bands shown in **Figure 5**.

Use information in **Figure 5** to:

- complete **Table 7**
- explain how you identified the child with CF.

You can use **F** to represent the dominant *CFTR* allele and **f** to represent the recessive F508del *CFTR* allele.

[3 marks]

Table 7

Lanes	Parent or child	Genotype
1		
2		
3		
4		

Explanation _____

_____

_____

_____

_____

Question 7 · information from earlier in the question

0 5 Leigh syndrome (LS) is a rare, recessive, inherited condition.

LS is caused by a mutation in any one of more than 75 different genes coding for proteins involved in oxidative phosphorylation.

In 80% of people with LS, these mutations occur in nuclear DNA. In 20% of people with LS, these mutations occur in mitochondrial DNA (mtDNA).

15% of the nuclear DNA mutations that cause LS occur in the *SURF1* gene. A mutated *SURF1* gene codes for a shorter polypeptide than a non-mutated *SURF1* gene.

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0 5 . 2 The population of the Faroe Islands is 49 053

Estimate the number of people in the Faroe Islands with LS caused by a mutation in the *SURF1* gene.

Use information in this question and the information on page 12.

Give your answer to the nearest whole number.

Show your working.

[3 marks]

Question 7 · A level Paper 3 · June 2024 · Q05.4 · 3 marks

0 5 . 4 LS usually causes death within the first three years of life.

Using all the information in this question, evaluate whether all people should be genetically screened for LS.

[3 marks]

Question 8 · information from earlier in the question

0 9 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.

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Question 8 · A level Paper 2 · June 2023 · Q09.4 · 2 marks

0 9 . 4 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.

[2 marks]

1 _____

_____

2 _____

_____

Question 9 · information from earlier in the question

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

Question 9 · A level Paper 2 · June 2018 · Q08.6 · 2 marks

0 8 . 6 The soybean cells were screened for the presence of the P34 protein. This process involved the use of gel electrophoresis to separate proteins extracted from soybean cells.

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

[2 marks]

1 _____

_____

_____

2 _____

_____

_____

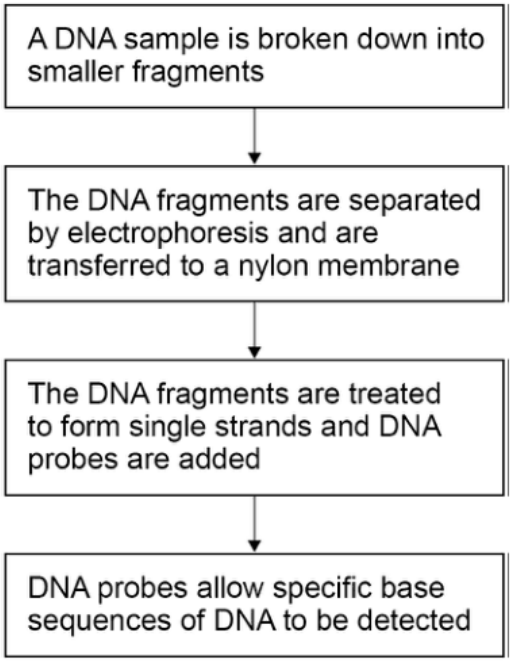
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Question 10 · information from earlier in the question

DNA probes are used to detect specific base sequences of DNA.

The process is shown in **Figure 6**.

Figure 6

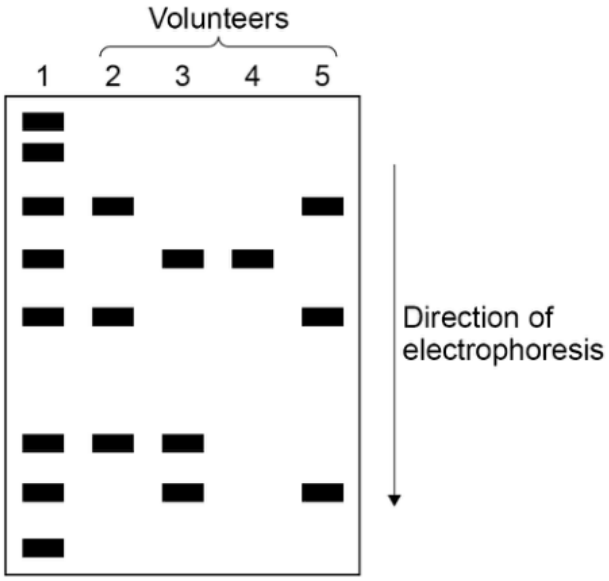


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A scientist used DNA probes and electrophoresis to screen four volunteers for five different viral DNA fragments.

Figure 7 shows the results the scientist obtained. The lanes numbered 2 to 5 represent the four volunteers.

Figure 7



0 8 . 4 Lane 1 of **Figure 7** enabled the size of the different viral fragments to be determined.

Suggest and explain how.

[2 marks]

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Question 11 · A level Paper 2 · June 2019 · Q08.5 · 1 mark

The lengths of the viral DNA fragments were:

- 600 base pairs
- 250 base pairs
- 535 base pairs
- 300 base pairs
- 500 base pairs.

0 8 . 5 Which volunteers had at least one of the viral DNA fragments with 250 base pairs or 535 base pairs?

[1 mark]

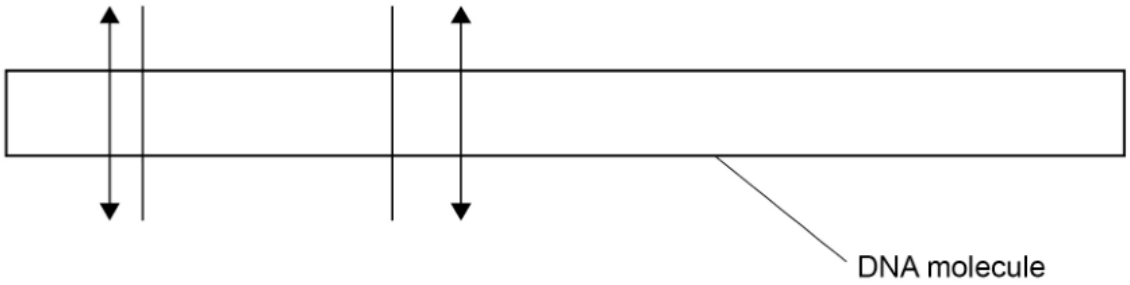
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Question 12 · information from earlier in the question

0 7 . 1 *Bam*H1 and *Hind*III are both restriction endonucleases.

Figure 4 shows the positions where these enzymes cut a linear molecule of DNA.

Figure 4



Key

Position DNA molecule cut by *Bam*H1

Position DNA molecule cut by *Hind*III

In two experiments, multiple copies of the DNA molecule shown in **Figure 4** 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 *Hind*III.

Complete **Figure 5** to show the relative positions of the bands following electrophoresis in experiments 1 and 2.

[2 marks]

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Question 12 · A level Paper 2 · June 2024 · Q07.2 · 2 marks

0 7 . 2 Suggest how you could determine the size of the different DNA fragments produced in these experiments.

[2 marks]

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Mark schemes

Question 1 · A level Paper 2 · June 2019 · Q08.1 · 2 marks

08.1	1. (Short) single strand of DNA; 2. Bases complementary (with DNA/allele/gene);	2

Question 2 · A level Paper 2 · June 2019 · Q08.3 · 1 mark

08.3	(So DNA) probe binds/attaches/anneals;	1	

Question 3 · A level Paper 3 · November 2021 · Q06.1 · 4 marks

06.1	1. Extract DNA and add restriction endonucleases/restriction enzymes; 2. Separate fragments using electrophoresis; 3. (Treat DNA to) form single strands **OR** (Treat DNA to) expose bases; 4. The probe will bind to/hybridise/base pair with the *SUT1*/gene; 5. Use autoradiography (to show the bound probe);	4 max

3. Ignore method used to separate strands

5. Accept use photographic or X ray film (to show the bound probe)

5. X rays alone is not sufficient

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Question 4 · A level Paper 2 · June 2017 · Q10.7 · 2 marks

10.7	1. (GCA/triplet) is common/found in other places; 2. Would not know if it was the mutation/allele/gene OR Produces 'false positives';	2	1. Accept: Probe will bind elsewhere.

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10.2	1. Use of PCR to amplify (DNA/sample); 2. Cut (DNA) using restriction endonuclease/enzymes; 3. Separate (DNA fragments) using electrophoresis; 4. Addition of (labelled) DNA probes **and** binding (by DNA hybridisation); 5. (Mutations) identified by fluorescence/radioactivity **OR** Compare positions/bands (to known) DNA sample with (all harmful) mutations;

4 max (3 x AO1, 1 x AO2)	1. Accept description of amplification. 3. Accept use of microarray for electrophoresis. 4. Ignore primers. 4. Reference to probe being complementary is insufficient. 5. Accept identification using X-

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09.3

1. and 2. **1 mark** per correct column;;

Lane	Parent or child	Genotype
1	Parent	Ff / fF
2	Child	ff
3	Parent	Ff / fF
4	Child	FF

3. (Lane 2 only contains) recessive/f allele which travels further due to fewer bases

OR

(Lane 2 only contains) recessive/f allele that travels further due to deletion of (3) bases;

3 (3 x AO3)

1 and 2. Accept if columns are reversed

2. Accept heterozygous, homozygous recessive, heterozygous and homozygous dominant

2. Accept use of different letters other than F and f

3. Accept 'triplet' for (3) bases

3. Accept F508del or mutation for recessive allele

3.In this instance accept gene for allele

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05.4	**2 max for mark point 1 to 4 OR 5 to 7** **Yes (no mark)** 1. Some people could be heterozygous/carriers; 2. Could prevent (human) suffering/death **OR** Could allow for (informed) decisions about having children; 3. (But only) in families/people with a history of LS **OR** (only) in families/people in the Faroe Islands (where high frequency/1: 1700); 4. Cost of screening might be cheaper than cost of treating LS; **No (no mark)** 5. It is rare (globally) **OR** (Only) 1 in 40 000 (globally); 6. Caused by (too) many genes/one of 75 genes **OR** Would need (too) many probes/75 probes; 7. (Too) expensive to produce tests/probes (for more than 75 different genes) **OR** (Too) expensive to screen **all**;	3 max (3 x AO3)

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Question 8 · A level Paper 2 · June 2023 · Q09.4 · 2 marks

09.4	1. Number of nucleotides/repeats/bases **OR** Length/mass; 2. (Negative) charge;	2 (2 x AO2)	1. Accept weight for mass. 1. Ignore 'short' on its own. 1. Accept number of base pairs. 1. Ignore density/size. 2. Accept 'polarity'. 2. Reject positive (charge).

Question 9 · A level Paper 2 · June 2018 · Q08.6 · 2 marks

08.6	1. Mass/number of amino acids/polypeptides; 2. Charge; 3. R groups (differ);	2 max	1. Accept weight for mass 1. Ignore density/size 1. Accept length of polypeptide/amino acid chain 1. Accept primary structure /sequence of amino acids. 1. Accept tertiary structure

Question 10 · A level Paper 2 · June 2019 · Q08.4 · 2 marks

08.4	1. (Lane 1 has DNA fragments) of known sizes/lengths; 2. Compare (position of viral fragment/s);	2

Question 11 · A level Paper 2 · June 2019 · Q08.5 · 1 mark

08.5	3, 4, 5 with these numbers in any sequence;	1	All three numbers required. Reject if more than three numbers given.

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Question 12 · A level Paper 2 · June 2024 · Q07.2 · 2 marks

07.2	1. (Separate) DNA fragments/ladder of known sizes/lengths; 2. Compare position/distance/bands with unknown fragment(s);	2 (2 x AO2)	1. Ignore mass

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