AQA – The control of gene expression – A2 Biology

1. June/2020/Paper_2/No.8

0 8 Alport syndrome (AS) is an inherited disorder that affects kidney glomeruli of both men and women. Affected individuals have proteinuria (high quantities of protein in their urine).

0 8 . 1 Suggest how AS could cause proteinuria.

[2 marks]

0 8 . 2 AS results from a sex-linked mutation.

In a male with AS, where would the sex-linked mutation be located?

Tick (✓) **one** box.

[1 mark]

- The homologous section of a Y chromosome ☐
- The homologous section of an X chromosome ☐
- The non-homologous section of a Y chromosome ☐
- The non-homologous section of an X chromosome ☐

Scientists investigated the use of transplanted stem cells to treat AS in mice.

The scientists set up four experimental groups.

Group **A** – 40 wild type* mice
Group **B** – 40 AS mice
Group **C** – 40 AS mice that received stem cells from AS mice
Group **D** – 40 AS mice that received stem cells from wild type mice

*Wild type mice are mice **not** affected by AS.

After 20 weeks, the scientists measured the quantity of protein in the urine using a scale from 0 (lowest quantity) to +++++ (highest quantity).

The results the scientists obtained are shown in **Table 2**.

Table 2

Group	Maximum quantity of protein in urine at 20 weeks	Percentage of mice with this quantity of protein
A	0	100
B	+++++	97.5
C	+++++	100
D	++	68

0 8 . 3 Using all the information, evaluate the use of stem cells to treat AS in humans.

[4 marks]

0 8 . 4 The scientists carried out further work to investigate how the transplanted stem cells developed after transplantation.

- The scientists transplanted stem cells from wild type male mice into AS female mice.
- After 20 weeks, they found that the quantity of protein in the urine of these female mice had significantly decreased.
- They examined cells from glomeruli in the female mice. Some of these cells contained a Y chromosome.

Suggest how the transplanted stem cells reduce proteinuria.

[2 marks]

2. June/2020/Paper_2/No.9

0 9 A scientist produced transgenic zebrafish.

She obtained a gene from silverside fish. The gene codes for a growth hormone (GH).

She inserted copies of this *GH* gene into plasmids. She then microinjected these recombinant plasmids into fertilised egg cells of zebrafish.

0 9 . 1 Describe how enzymes could be used to insert the *GH* gene into a plasmid.

[2 marks]

0 9 . 2 Microinjection of DNA into fertilised egg cells is a frequent method of producing transgenic fish. However, the insertion of the transferred gene into nuclear DNA may be delayed. Consequently, the offspring of transgenic fish may not possess the desired characteristic.

Suggest and explain how delayed insertion of the *GH* gene could produce offspring of transgenic fish without the desired characteristic.

[2 marks]

The scientist investigated whether the transferred *GH* gene increased the growth of transgenic zebrafish. She microinjected 2000 fertilised egg cells with the *GH* plasmid and left 2000 fertilised egg cells untreated. After 12 months, she determined the mean mass of the transgenic and non-transgenic fish.

The results the scientist obtained are shown in **Table 3**.

Table 3

A value of ±2 × SD from the mean includes over 95% of the data.

Type of zebrafish	Mean mass of zebrafish / g (± 2 × SD)
Transgenic	1.79 (± 0.37)
Non-transgenic	0.68 (± 0.13)

0 9 . 3 Using **Table 3**, what can you conclude about the effectiveness of the *GH* gene on the growth of zebrafish?

[2 marks]

0 9 . 4 Explain how **two** features of the design of this investigation helped to ensure the validity of any conclusions obtained.

Do **not** include calculating the mean or SD in your answer.

[2 marks]

1

2

3. June/2019/Paper_2/No.8

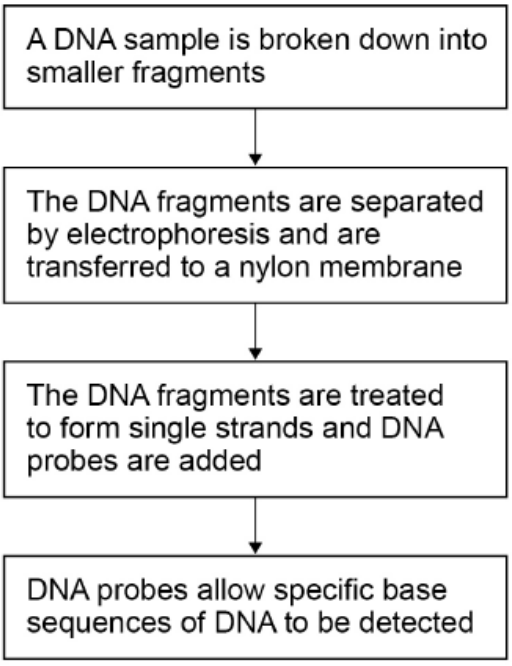
0 8 . 1 What is a DNA probe?

[2 marks]

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

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

Figure 6



0 8 . 2 Describe how the DNA is broken down into smaller fragments.

[2 marks]

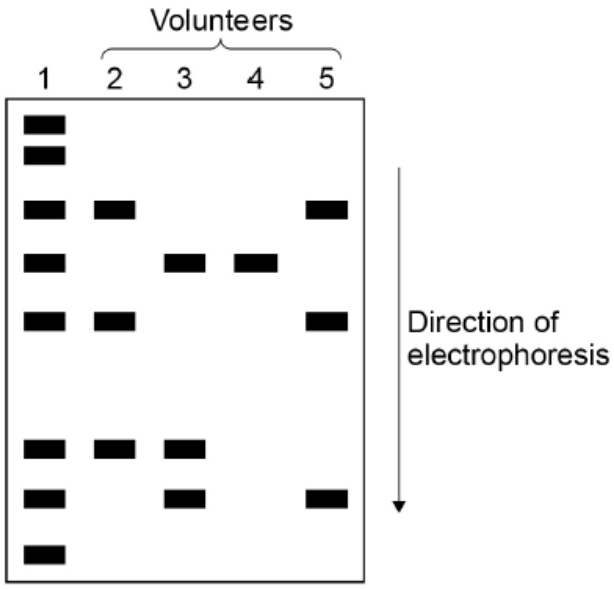
0 8 . 3 The DNA on the nylon membrane is treated to form single strands. Explain why.

[1 mark]

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]

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]

4. June/2019/Paper_2/No.10

1 0 Guillain–Barré syndrome is a rare disease in which the immune system damages the myelin sheath of neurones. Myelin sheath damage can cause a range of symptoms, for example numbness, muscular weakness and muscular paralysis. Sometimes, neurones of the autonomic nervous system are affected, causing heart rate irregularities.

Huntington's disease is a disorder caused when a protein called huntingtin damages the brain. Huntingtin is produced because of a dominant, mutant allele.

The first successful drug trial to reduce concentrations of huntingtin in the human brain involved 46 patients. The patients received the drug for 4 months. The concentration of huntingtin was reduced in all the patients. The drug was injected at the base of the spine into the cerebrospinal fluid bathing the brain and spinal cord. The drug contains single-stranded DNA molecules. These single-stranded molecules inhibit the mRNA needed to produce huntingtin.

Symptoms of Huntington's disease can start at any time, but usually develop between 30 and 50 years of age. The likelihood and age when symptoms start are linked to the number of CAG base sequence repeats in the gene for Huntington's disease. However, recent studies have suggested that epigenetics may also affect the age when symptoms first start.

1 0 . 1 Damage to the myelin sheath of neurones can cause muscular paralysis (lines 2–4).

Explain how.

[3 marks]

1 0 . 2 Sometimes Guillain–Barré syndrome causes heart rate irregularities (lines 4–5).

Suggest and explain why.

[3 marks]

1 0 . 3 The first successful drug trial to reduce concentrations of huntingtin in the brain used single-stranded DNA molecules (lines 13–14).

Suggest and explain how this drug could cause a reduction in the concentration of the protein huntingtin.

[3 marks]

1 0 . 4 Scientists from the first successful drug trial to reduce concentrations of huntingtin (lines 9–11) reported that the drug is not a cure for Huntington's disease.

Suggest **two** reasons why the drug should **not** be considered a cure.
Do **not** include repeats of the drug trial in your answer.

[2 marks]

1 _____

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2 _____

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1 0 . 5 Suggest **two** reasons why people had the drug injected into the cerebrospinal fluid (lines 12–13) rather than taking a pill containing the drug.

[2 marks]

1 _____

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2 _____

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1 0 . 6 Suggest and explain **one** way epigenetics may affect the age when symptoms of Huntington's disease start.

[2 marks]

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