Q1.

Sickle cell disease (SCD) is a group of inherited disorders. People with SCD have sickle-shaped red blood cells. A single base substitution mutation can cause one type of SCD. This mutation causes a change in the structure of the beta polypeptide chains in haemoglobin.

(a) Explain how a single base substitution causes a change in the structure of this polypeptide.

Do **not** include details of transcription and translation in your answer.

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(3)

Q2.

Mitochondrial DNA (mtDNA) is a small circular DNA molecule located in mitochondria. It is 16 569 nucleotides long and contains 37 genes and a control region.

Sports scientists investigated whether a mutation in the control region of mtDNA in human males was related to an ability to exercise for longer.

- The males in Group **T** had thymine at nucleotide position 16 519
- The males in Group **C** had a mutation resulting in cytosine at nucleotide position 16 519

(a) The control regions of Group **T** and Group **C** were the same length.

Name the type of gene mutation that is most likely to have occurred at nucleotide position 16 519

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(1)

Group **T** and Group **C** completed the same 8-week training programme. The following measurements were taken at the start of the 8-week programme, and again at the end.

1. $VO_{2\ max}$ (a measure of maximal oxygen uptake).
2. Citrate synthase (CS) activity (CS is an enzyme involved in the Krebs cycle).

The scientists then calculated the percentage increase in each measurement in both groups.

Figure 1 and **Figure 2** show their results.

Figure 1

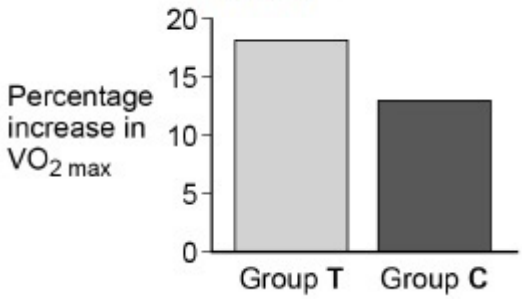
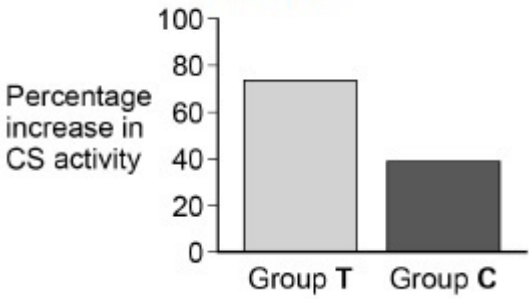


Figure 2



(b) A student concluded from **Figure 1** and **Figure 2** that training has a positive effect on $VO_{2\ max}$ and CS activity.

Evaluate the student's conclusion.

(3)

(c) The mitochondrial DNA (mtDNA) control region is an area of mtDNA that is non-coding. This region stimulates the synthesis of both mtDNA and mitochondrial messenger RNA.

Use this information to suggest **two** reasons why the mutation at nucleotide position 16 519 could lead to the differences seen in **Figure 2**.

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2 _____

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(2)

Q3.

Some autism spectrum disorders (ASDs) are associated with a mutation affecting the neuroligin-3 gene. This gene codes for a protein called NL3, that is found in synapses.

Scientists investigated the effects of a mutation affecting NL3 in mice. They obtained brains from mice with the mutation and from mice without the mutation. For each type of mouse they:

- obtained a solution containing all of the proteins from synapses in one part of the brain
- separated these proteins using gel electrophoresis
- identified and measured the amount of three proteins from the solution using three different labelled antibodies.

The three proteins are parts of a postsynaptic membrane receptor.

The diagram below shows the scientists' results. Each band shows the presence of a protein. The size of a band shows the amount of the protein present.

(a) The mutation affecting NL3 in these mice was a substitution in the neuroligin-3 gene.

What is a substitution mutation?

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(1)

(d) What do these data show about the effects of the mutation on the proteins?

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(2)

(e) These proteins are part of a receptor found in synapses in the part of the brain called the hippocampus. A high ratio of NR2B to NR2A protein in this receptor has been associated with good memory.

Using all of the information, suggest how the mutation affecting the NL3 protein may affect a mouse.

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(2)

(Total 10 marks)