A-Level Biology

Gene Mutations

Question Paper

Time available: 56 minutes
Marks available: 41 marks

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1. (a) There are different types of gene mutation.

Put a tick (✓) in the box next to the statement which describes **incorrectly** the effect of the mutation in an exon of a gene.

A substitution may not result in a change to the encoded amino acid.	☐
An inversion will result in a change in the number of DNA bases.	☐
A deletion will result in a frame shift.	☐
An addition will result in a frame shift.	☐

(1)

(b) Describe how alterations to tumour suppressor genes can lead to the development of tumours.

(3)

(c) A type of malignant tumour cell divides every 8 hours.

Starting with one of these cells, how many tumour cells will be present after 4 weeks? Assume none of these cells will die.

Give your answer in standard form.

Answer = _____

(2)

(Total 6 marks)

2. 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)

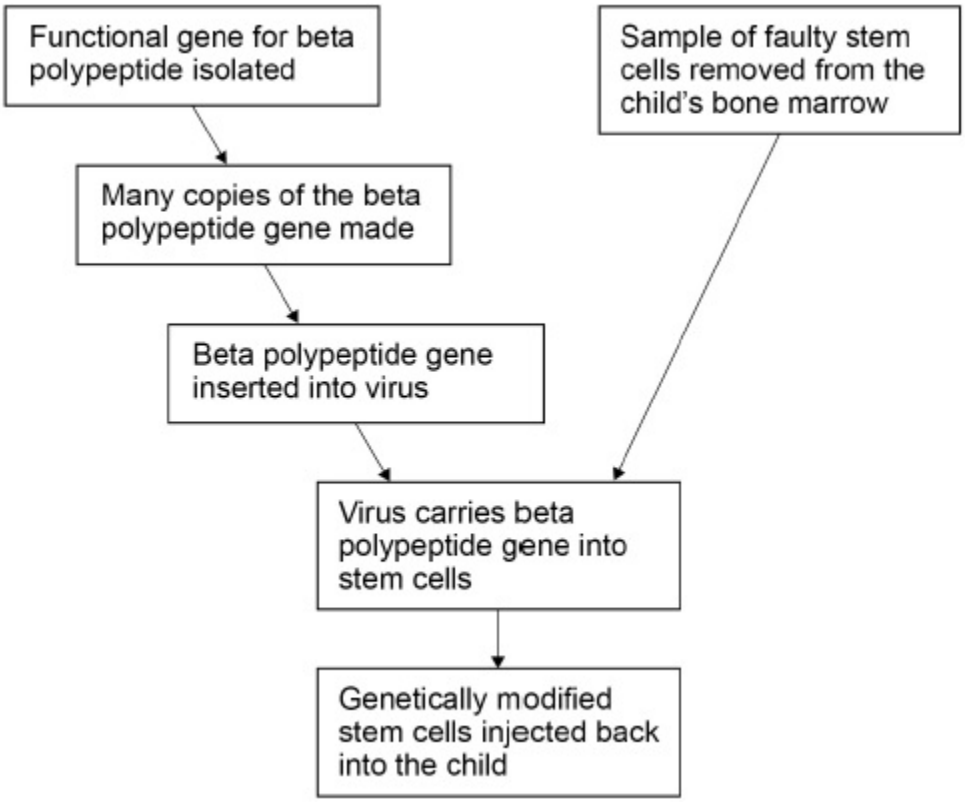
Haematopoietic stem cell transplantation (HSCT) is a long-term treatment for SCD. In HSCT, the patient receives stem cells from the bone marrow of a person who does not have SCD. The donor is often the patient's brother or sister. Before the treatment starts, the patient's faulty bone marrow cells have to be destroyed.

(b) Use this information to explain how HSCT is an effective long-term treatment for SCD.

(3)

A new long-term treatment for SCD involves the use of gene therapy.

The diagram shows some of the stages involved in this treatment in a child with SCD.



(c) Some scientists have concluded that this method of gene therapy will be a more effective long-term treatment for SCD than HSCT. Use all the information provided to evaluate this conclusion.

(3)

(Total 9 marks)

3. Phenylketonuria is a disease caused by mutations of the gene coding for the enzyme PAH. The table shows part of the DNA base sequence coding for PAH. It also shows a mutation of this sequence which leads to the production of non-functioning PAH.

DNA base sequence coding for PAH	C	A	G	T	T	C	G	C	T	A	C	G
DNA base sequence coding for non-functioning PAH	C	A	G	T	T	C	C	C	T	A	C	G

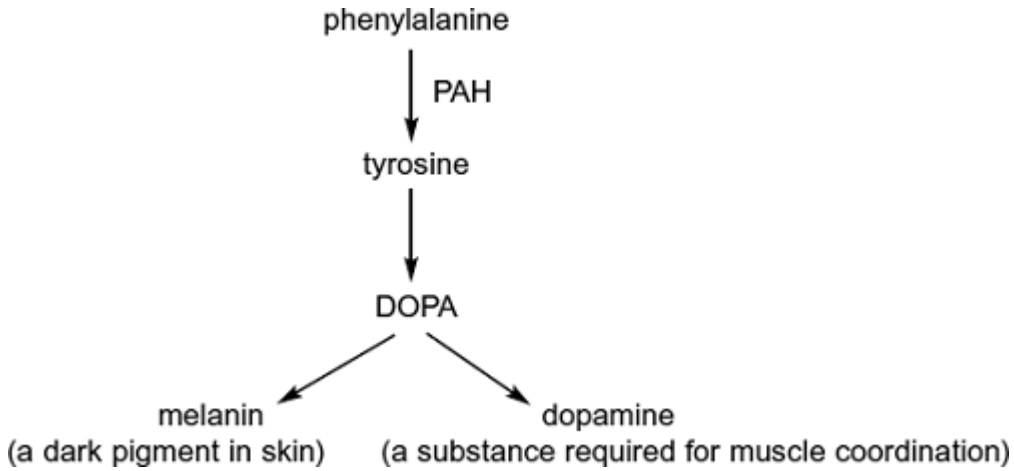
(a) (i) What is the maximum number of amino acids for which this base sequence could code?

(1)

(ii) Explain how this mutation leads to the formation of non-functioning PAH.

(3)

PAH catalyses a reaction at the start of two enzyme-controlled pathways.
The diagram shows these pathways.



(b) Use the information in the diagram to give **two** symptoms you might expect to be visible in a person who produces non-functioning PAH.

1. _____

2. _____

(2)

(c) One mutation causing phenylketonuria was originally only found in one population in central Asia. It is now found in many different populations across Asia. Suggest how the spread of this mutation may have occurred.

_____

_____

_____

(1)

(Total 7 marks)

4. 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

_____

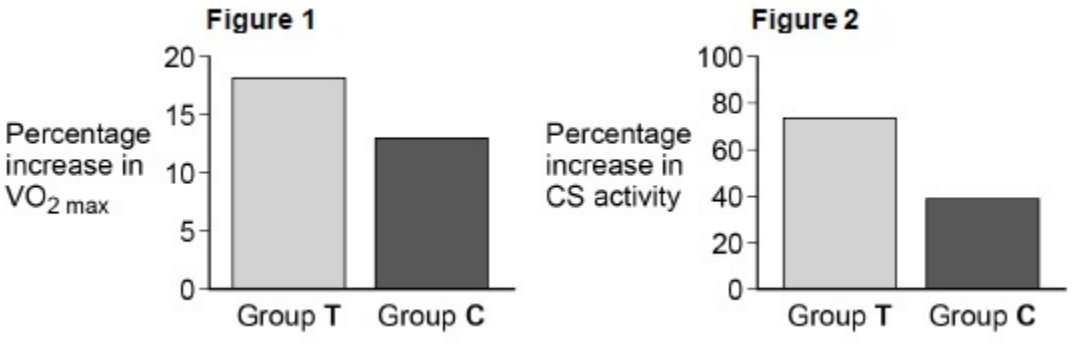
(1)

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.



(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**.

1 _____

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2 _____

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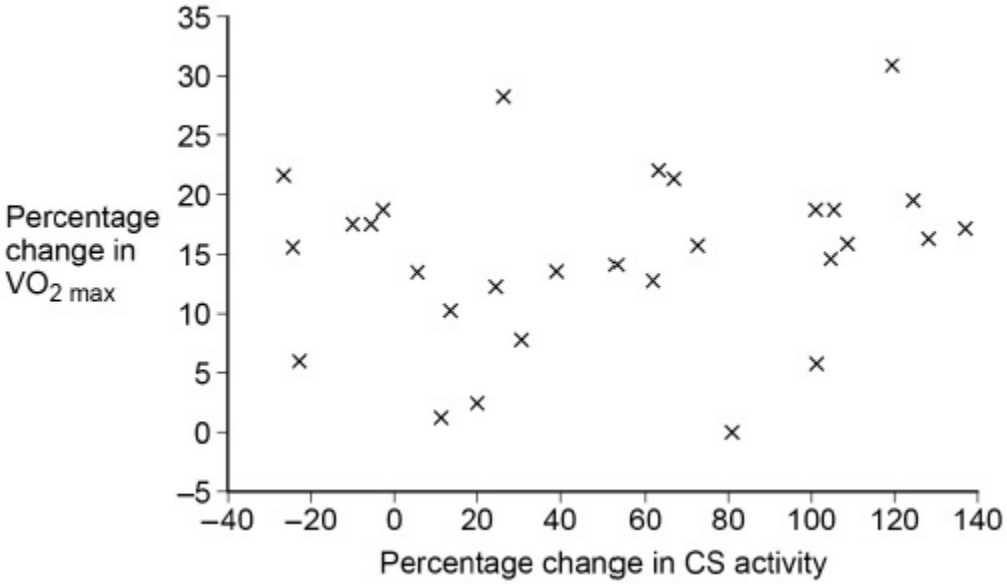
(2)

The sports scientists investigated whether there was a correlation between the percentage change in $VO_{2\ max}$ and percentage change in CS activity in Group **T**.

Figure 3 shows their results.

Figure 3



(d) ‘Having thymine at nucleotide position 16 519 in Group **T** causes an increase in ability to exercise for longer.’

Evaluate this conclusion.

Use **all** the data in this question.

(3)

(Total 9 marks)

5. 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?

_____

_____

_____

(1)

(b) Suggest how gel electrophoresis separated the proteins obtained from the synapses.

_____

_____

_____

_____

_____

(2)

(c) Each type of labelled antibody binds specifically to one of the proteins.

Explain why.

(3)

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

(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.

(2)

(Total 10 marks)