

**SMART EXAM RESOURCES**  
**TOPIC QUESTIONS: NUCLEIC ACID AND PROTEIN**  
**SYNTHESIS**  
**SUB-TOPIC: GENE MUTATION**  
**SET-1-QP-MS**

**1** The gene coding for glycogen synthase in muscle cells is known as *GYS1*.

There are a number of known mutations for *GYS1*.

Outline how a mutation in *GYS1* can lead to the formation of an altered polypeptide where one amino acid is replaced by a different amino acid.

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.....[3]

## MARK SCHEME:

- 1 (in DNA/gene) altered, sequence / AW, of, nucleotides / bases ;  
I DNA sequence
- 2 base substitution  
or base / nucleotide, replaces another, base / nucleotide;  
A example *must be in context of, DNA/gene*
- 3 (mRNA synthesised) during transcription ;
- 4 (mutation leads to) altered / AW, mRNA / messenger RNA ;
- 5 (only) one (mRNA) codon changed / a different codon ;  
A one DNA, triplet / codon, changed I *ref. to codons* changed
- 6 tRNA, with / has, a different anticodon ;
- 7 (tRNA) brings, a different / a changed / the incorrect, amino acid, during  
translation / to the ribosome ;
- 8 codon-anticodon, binding / complementary / AW ; A matches  
R amino acid with anticodon

[max 3]

**2**

Outline how mutations can cause healthy cells to become tumour cells.

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.....[3]

**MARK SCHEME:**

(a) (i) GTG ;

ACU ;

leu ;

[3]

(ii) primary structure ;

[1]

# 3

In sickle cell anaemia, the amino acid at position 6 in the chain is valine and not glutamic acid.

Explain how a single change in the DNA triplet for the sixth amino acid of the gene coding for the  $\beta$  chain leads to the production of a different amino acid sequence.

.....[5]

## MARK SCHEME:

- 1 mutation ;
- 2 base substitution/T → A in template strand of DNA/AW ;

### *transcription*

- 3 DNA has CAC as 6<sup>th</sup> triplet ;
  - 4 (so) mRNA has GUG as (6<sup>th</sup>) codon ;
- allow one mark for altered mRNA codon if no marks gained for mps 3 and 4*

### *translation*

- 5 different tRNA involved/tRNA specific to val and not glu ;
  - 6 anticodon on tRNA has CAC (with valine) ;
  - 7 tRNA brings, incorrect amino acid/val, to ribosome ;
  - 8 further detail ; e.g. incorrect amino acid incorporated into growing polypeptide chain
- [max 5]