THIS IS A LEGACY SPECIFICATION

OCR
RECOGNISING ACHIEVEMENT

ADVANCED SUBSIDIARY GCE

HUMAN BIOLOGY

2858/01

Case Studies

OCE/T59940

Candidates answer on the question paper

OCR Supplied Materials:
- Insert (inserted)

Other Materials Required:
- Electronic calculator
- Ruler (cm/mm)

Monday 1 June 2009
Afternoon

Duration: 45 minutes

2858012

Candidate Forename		Candidate Surname	

Centre Number						Candidate Number				

INSTRUCTIONS TO CANDIDATES

- Write your name clearly in capital letters, your Centre Number and Candidate Number in the boxes above.
- Use black ink. Pencil may be used for graphs and diagrams only.
- Read each question carefully and make sure that you know what you have to do before starting your answer.
- Answer **all** the questions.
- Do **not** write in the bar codes.
- Write your answer to each question in the space provided, however additional paper may be used if necessary.

INFORMATION FOR CANDIDATES

- The number of marks is given in brackets **[ ]** at the end of each question or part question.
- The total number of marks for this paper is **45**.
- You may use an electronic calculator.
- You are advised to show all the steps in any calculations.
- This document consists of **12** pages. Any blank pages are indicated.

FOR EXAMINER'S USE		
Qu.	**Max.**	**Mark**
1	**23**	
2	**22**	
TOTAL	**45**	

Answer **all** the questions.

1 This question is based on the article **'THALASSAEMIA – NOT KNOWING WHEN TO STOP!' (Case Study 1)**.

(a) In the case study, you are given the names of several amino acids that occupy different positions in the alpha or beta chains of the protein haemoglobin.

Fig. 1.1 is a diagram of the amino acid alanine.

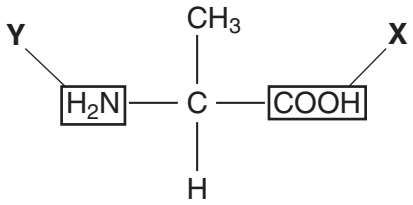

Fig. 1.1

(i) Name the parts labelled **X** and **Y**.

X .....

Y ..... **[2]**

(ii) **On Fig. 1.1, draw a circle** around the part of the molecule that will vary for different amino acids. **[1]**

(b) Complete the following passage about protein structure by inserting the most appropriate word or words.

The sequence of amino acids in a protein is known as the primary structure. The covalent bonds that join the amino acids together are called ..... bonds, and the order in which the amino acids join up is laid down by the genetic code on the ..... strand of the DNA in the nucleus. The chain of amino acids can be coiled into a secondary structure such as ..... , and this structure is stabilised by ..... bonds. The chain can be further coiled and folded into a specific shape, which is stabilised by bonds between ..... of the amino acids. In the haemoglobin molecule there are ..... chains, which together form the ..... structure of this protein. **[7]**

(c) You are told in the case study that some mutations in the gene for the beta haemoglobin chain can result in different amino acids being found at the 59th position in the chain.

Explain how a mutation in the DNA could result in **no change** to the amino acid at this position.

.....

.....

.....

.....

.....

..... **[2]**

(d) You are told in the case study that some mutations can result in haemoglobin having a 'higher affinity' for oxygen than normal adult haemoglobin.

Conditions where the affinity of haemoglobin for oxygen is higher than normal can result in more oxyhaemoglobin being present in the blood cells returning to the heart from tissues such as muscle.

(i) Suggest what effect the presence of more oxyhaemoglobin in the red blood cells will have on the **colour** of the blood in veins?

..... **[1]**

(ii) Suggest what would happen if blood entering active muscle tissue cannot release oxygen from oxyhaemoglobin.

.....

.....

.....

.....

.....

..... **[3]**

(e) A patient with Constant Spring (CS) thalassaemia is found to have a red blood cell count of 4.4×10^9 cells cm^{-3}.

Outline how a haemocytometer could be used to obtain a red blood cell count, such as the one given above.

.....

.....

.....

.....

.....

.....

.....

.....

.....

.....

.....

.....

.....

.....

.....

.....

..... **[4]**

(f) You are told in the case study that CS thalassaemia results from a mutation in the gene for the alpha haemoglobin chain. This results in a stop codon becoming a codon that would code for an amino acid.

UGA is an example of a stop codon.

Table 1.1 shows some **DNA triplets** and their corresponding amino acids.

Table 1.1

DNA triplet	amino acid
ATA	tyrosine
ACC	tryptophan
ATG	tyrosine
TTC	lysine

Using the information in Table 1.1, explain how a **point mutation** could result in changing a stop codon into a codon that codes for an amino acid.

.....

.....

.....

.....

.....

..... **[3]**

[Total: 23]

2 This question is based on the article '**THE OTHER PHILADELPHIA STORY' (Case Study 2)**.

(a) You are told in the case study that some haematologists work in transfusion laboratories. The haematologist will need to know which blood groups each sample of blood belongs to.

Explain what is meant by a *blood group*.

.....

.....

.....

.....

.....

.....

.....

.....

..... **[3]**

(b) You are told in the case study that chronic myeloid leukaemia (CML) can result in the failure of cells to differentiate into granulocytes, such as neutrophils, and into macrophages.

With reference to the development of neutrophils, explain what is meant by the term *differentiate*.

.....

.....

.....

.....

.....

..... **[3]**

(c) In the case study, Trish refers to the cell cycle.

(i) State **one** process that happens **in the nucleus** of cells during interphase of the cell cycle.

..... **[1]**

(ii) Describe the appearance of a chromosome during metaphase. You may use a labelled diagram.

.....

.....

.....

.....

[2]

(iii) Explain why CML is **not** an inherited disease.

.....

.....

.....

.....

..... **[2]**

(d) In the case study, you are told that a drug inhibits an enzyme in the CML cells. This drug stops the CML cells dividing uncontrollably.

Explain how enzyme inhibitors work.

....

....

....

....

....

....

....

....

....

....

....

....

....

....

....

....

....

....

....

....

....

....

....

....

.... **[7]**

(e) You are told in the case study that 84% of patients in one trial showed no sign of the Philadelphia chromosome after treatment.

If the number of patients involved in the trial was 312, calculate how many patients showed no sign of the Philadelphia chromosome.

Show your working.

Answer = ... **[2]**

(f) You are told in the case study that patients who respond to the drug have to keep taking it for the rest of their lives.

Comment on the dilemma faced by Primary Care Trusts (PCTs) in providing long-term drug treatment for conditions such as CML.

.....

.....

.....

.....

..... **[2]**

[Total: 22]

END OF QUESTION PAPER

BLANK PAGE

PLEASE DO NOT WRITE ON THIS PAGE

BLANK PAGE

PLEASE DO NOT WRITE ON THIS PAGE

PLEASE DO NOT WRITE ON THIS PAGE