1 Cystic fibrosis is a genetic disease caused by mutations in the CFTR gene.
This disease can be classified according to the effect of the different gene mutations on the CFTR protein.

The table below shows the classification of cystic fibrosis.

Class	Effect on the CFTR protein
I	CFTR protein is not synthesised.
II	CFTR protein is mis-folded and is not found in the correct location.
III	CFTR protein is mis-folded and is found in the correct location, but does not function properly.
IV	CFTR protein has a faulty opening.
V	CFTR protein is synthesised in smaller quantities than normal.
VI	CFTR protein breaks down quickly after it is synthesised.

(a) For class I cystic fibrosis, suggest how a mutation in the CFTR gene could result in no CFTR protein being synthesised.

(2)

(b) Class II cystic fibrosis results from the CFTR protein being located in the wrong place.

Describe the correct location for the CFTR protein.

(2)

(c) The mutation causing class III cystic fibrosis results in a change in the primary structure of the CFTR protein.

Explain why this would result in the CFTR protein being mis-folded.

(2)

(d) For class IV cystic fibrosis, explain why a faulty opening of the CFTR protein would affect the functioning of this protein.

(2)

(e) For a person with class V cystic fibrosis, describe the effect of having smaller quantities of CFTR protein.

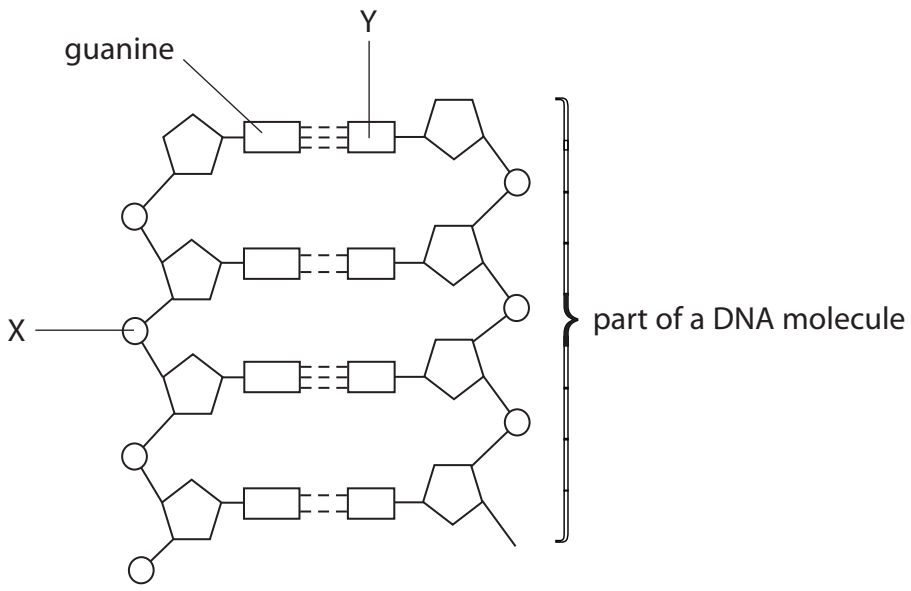
(2)

(f) For class VI cystic fibrosis, suggest how the CFTR protein is broken down.

(2)

(Total for Question 1 = 12 marks)

2 (a) The diagram below shows part of a DNA molecule.



(i) Place a cross ☒ in the box next to the molecule represented by the letter X

(1)

☒ **A** Deoxyribose

☒ **B** Phosphate

☒ **C** Ribose

☒ **D** Uracil

(ii) Place a cross ☒ in the box next to the molecule represented by the letter Y

(1)

☒ **A** Adenine

☒ **B** Cytosine

☒ **C** Thymine

☒ **D** Uracil

(iii) Place a cross ☒ in the box next to the name of the bonds holding the two strands of DNA together.

(1)

☒ **A** Ester bonds

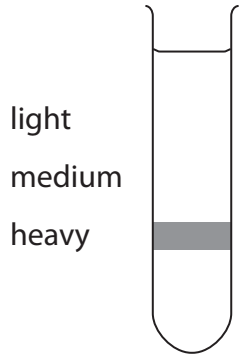
☒ **B** Glycosidic bonds

☒ **C** Hydrogen bonds

☒ **D** Peptide bonds

(b) A culture of bacteria had its DNA labelled with the heavy isotope of nitrogen (^{15}N).

The diagram below shows the position of the DNA band in the centrifuge tube when the DNA was labelled with the heavy isotope of nitrogen, ^{15}N.

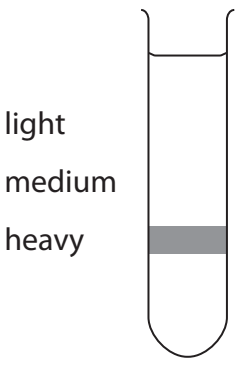


The bacterial culture was then allowed to reproduce using nucleotides containing the normal isotope of nitrogen (^{14}N).

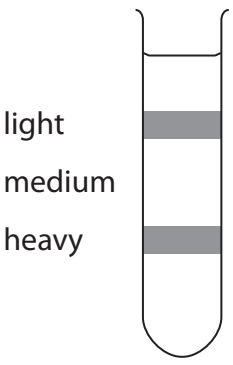
(i) Place a cross ☒ in the box below next to the tube showing the correct pattern of DNA after the bacteria have divided once.

(1)

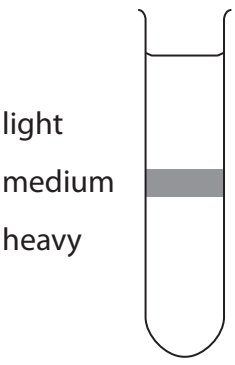
A ☒



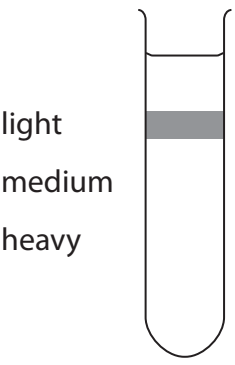
B ☒



C ☒

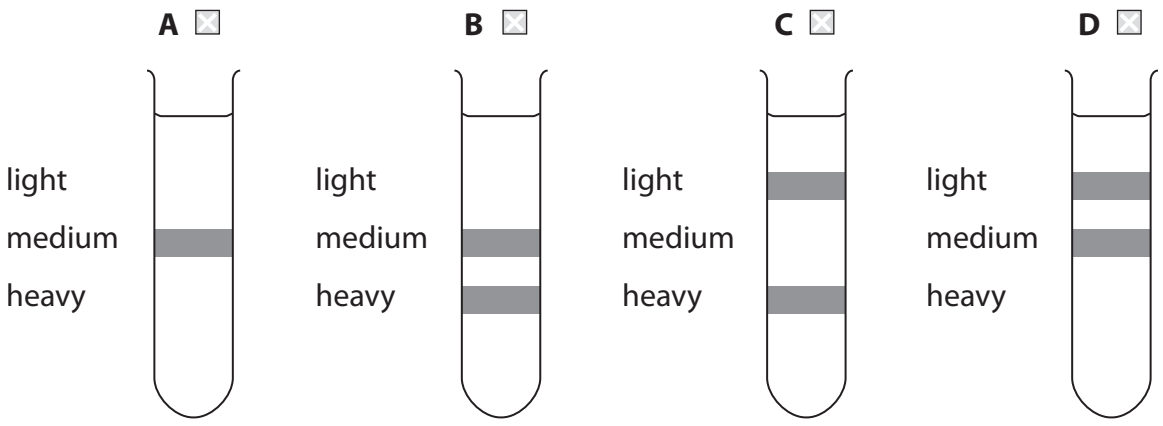


D ☒



(ii) Place a cross ☒ in the box below next to the tube showing the correct pattern of DNA after the bacteria have divided twice.

(1)



(c) Name the place in a eukaryotic cell where messenger RNA will be synthesised.

(1)

.....

(d) Achondroplasia is an inherited condition that results in restricted growth in humans. This condition is caused by a dominant allele (A).

Fetuses which are homozygous for the allele for achondroplasia are rarely born alive.

Two parents who both have achondroplasia would like to have children. They are concerned about the risk of their child inheriting two dominant alleles and dying before birth.

(i) Describe **one** advantage and **one** disadvantage to these parents of genetic screening of their fetus.

(2)

Advantage: .....

.....

.....

.....

Disadvantage: .....

.....

.....

.....

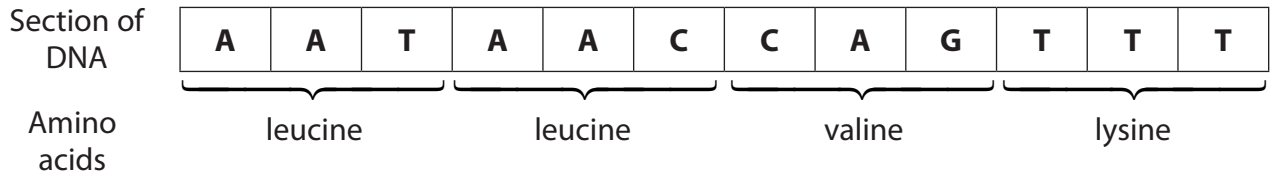
(ii) In the space below, draw a suitable genetic diagram to show the probability of a child from these parents growing up without achondroplasia.

(4)

probability.....

(Total for Question 2 = 12 marks)

3 The diagram below shows the base sequence on a short section of DNA consisting of 12 mononucleotides. This base sequence contains the genetic code for a short section in the primary structure of a polypeptide.



(a) Name each of the bases represented by the letters, **A**, **C**, **G** and **T** in the diagram.

(1)

A .....

C .....

G .....

T .....

(b) Using the sequence shown in the diagram, explain the meaning of each of the following terms.

(i) Triplet code

(2)

.....

.....

.....

.....

(ii) Non-overlapping

(2)

.....

.....

.....

.....

(iii) Degenerate

(2)

(c) Place a cross ☒ in the box next to the names of the two components, other than the bases, that form part of each mononucleotide in this sequence.

(1)

☒ **A** deoxyribose and nitrate

☒ **B** deoxyribose and phosphate

☒ **C** ribose and nitrate

☒ **D** ribose and phosphate

*(d) Transcription of this section of DNA forms a complementary strand of mRNA.

Describe how translation of this mRNA synthesises part of a polypeptide molecule.

(5)

(Total for Question 3 = 13 marks)

4 The sequence of bases in DNA determines the sequence of amino acids in a polypeptide.

The table below shows the genetic code for each amino acid.

TTT TTC	Phe	TCT TCC TCA TCG	Ser	TAT TAC	Tyr	TGT TGC	Cys
TTA TTG	Leu			TAA TAG	Stop	TGA	Stop
						TGG	Trp
CTT CTC CTA CTG	Ieu	CCT CCC CCA CCG	Pro	CAT CAC	His	CGT CGC CGA CGG	Arg
				CAA CAG	Gln		
ATT ATC ATA	Ile	ACT ACC ACA ACG	Thr	AAT AAC	Asn	AGT AGC	Ser
				AAA AAG	Lys	AGA AGG	Arg
ATG	Met						
GTT GTC GTA GTG	Val	GCT GCC GCA GCG	Ala	GAT GAC	Asp	GGT GGC GGA GGG	Gly
				GAA GAG	Glu		

(a) The diagram below shows the DNA base sequence coding for part of a polypeptide.

A	T	G	G	G	C	A	T	T

(i) Using the information in the table, state the order of amino acids for this part of the polypeptide.

(1)

.....

(ii) Explain what is meant by the term **non-overlapping genetic code**.

(1)

.....

.....

(b) (i) Explain why there are **three** bases in each of the codes shown in the table.

(2)

.....

.....

.....

(ii) Suggest an advantage for most amino acids having more than one code. Give an explanation for your answer.

(2)

(c) Explain the role of the base sequences TAA, TAG and TGA.

(4)

(d) Explain how the amino acids are joined together in a polypeptide.

(3)

(Total for Question 4 = 13 marks)