

OCR A-LEVEL BIOLOGY B • H422

BIOLOGY



EXAM QUESTION PACK

2.1.4 NUCLEIC ACIDS

SCORE

/133

PERCENTAGE

%

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1(a) Nucleic acids are required for the synthesis of proteins.

For each of the nucleic acids below, describe its role in protein synthesis.

(i) Messenger RNA (mRNA)

----- [1]

(ii) Transfer RNA (tRNA)

----- [1]

(b) DNA is the nucleic acid that provides the genetic information required for protein synthesis. This information is known as the genetic code.

Complete the passage below about the nature of the genetic code using the most appropriate word(s).

The sequence of bases in one strand of DNA provides the genetic code for making proteins.

Sections of DNA that code for specific polypeptides are known as

A sequence of nucleotide bases is required to form a DNA codon

that codes for an amino acid. However, the genetic code is, which

means that there is more than one codon for each amino acid. Each DNA codon is read

separately which means that there is no of bases in the sequence

during transcription.

[4]

2 Spermatogenesis in human males produces haploid gametes, called spermatozoa (sperm cells).

During spermatogenesis, the chromatin in the nuclei of sperm cells undergoes a complex transition where histone is replaced by another protein, protamine, which binds to the DNA.

(i) Protamine contains large quantities of the polar amino acid, arginine.

Suggest why having large quantities of arginine enables protamine to bind effectively to DNA molecules.

[2]

(ii) Substituting histone with protamine allows DNA to be more densely packaged.

Suggest **one** advantage of having densely packaged DNA in a sperm nucleus.

[1]

3(a) Please refer to the Advance Notice article ‘Mitochondrial DNA and mitochondrial disease’ in the Insert for QP H422/02, November 2020.

Mitochondrial DNA (mtDNA) is much smaller than nuclear DNA and is similar to prokaryotic DNA.

(i) Give **two** ways, apart from size, in which mtDNA differs from nuclear DNA.

1. -----

2. -----

[2]

(ii) Complete the following table of comparisons between mtDNA, RNA and ATP. Put a tick (✓) in each box where that component is present.

The first line has been completed for you.

Component	mtDNA	RNA	ATP
adenine	✓	✓	✓
ribose			
uracil			
pyrimidine			

[3]

(iii) The mitochondrial genome contains 37 genes. Of these, only 13 code for proteins. Suggest what the remaining genes encode.

[1]

(b) Like replication of nuclear DNA, replication of mtDNA is semi-conservative.

(i) Explain what is meant by the term semi-conservative replication.

----- [2]

(ii) Explain how mutations in the catalytic and exonuclease regions of the *POLG* gene could have different effects on the replication of mtDNA.

----- [2]

- 4 DNA normally contains four different nucleotides. Scientists have managed to create cells with six different nucleotides in their DNA.

The two new nucleotides are known as nucleotides X and Y.

Fig. 1.1 shows a section of a DNA molecule containing all six nucleotides, including X and Y.

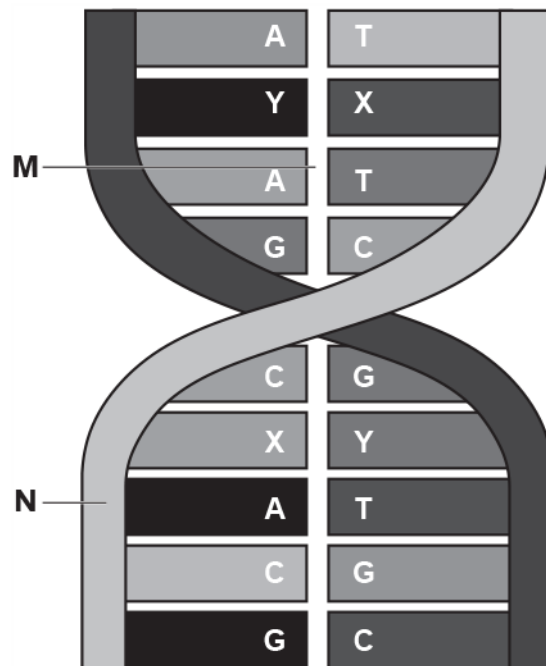


Fig. 1.1

- (i) Name the bonds found in the region labelled M on Fig. 1.1.

----- [1]

- (ii) Name the **two** components of DNA found in the region labelled N on Fig. 1.1.

----- [1]

5 Chromosome 1 in humans is composed of 2.5×10^9 base pairs.

DNA polymerase has an error rate of 1 base per 10^8 bases.

Which of the options, **A** to **D**, is the theoretical number of incorrect bases incorporated by DNA polymerase during replication of chromosome 1?

A 2.5

B 5

C 25

D 50

Your answer

[1]

- (b) The actions of CRISPR are mediated by an enzyme-RNA complex. The enzyme, Cas9, is attached to a guide RNA molecule, as shown in Fig. 35.

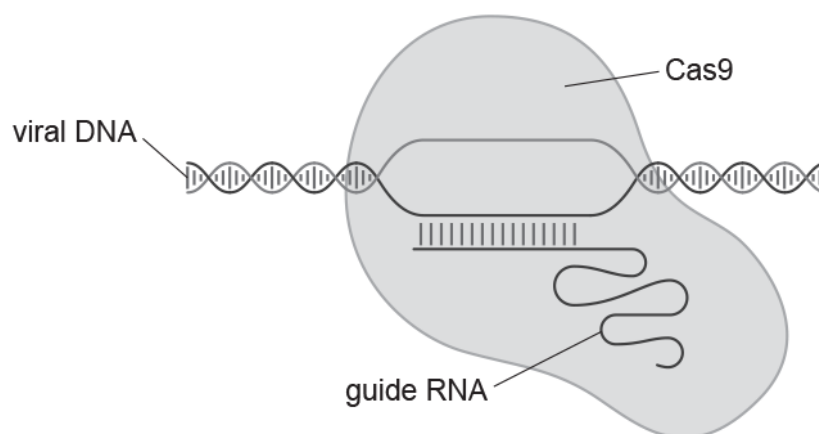


Fig. 35

The guide RNA molecule is complementary to the viral DNA molecule. Upon binding of the guide RNA to viral DNA, Cas9 cuts straight through both strands of the DNA at a precise position. The gene encoded by the DNA is inactivated. This prevents the virus from replicating inside the bacterial cell.

- (i) Cas9 is an endonuclease enzyme because it cuts within a nucleotide sequence.

Name another type of bacterial endonuclease enzyme.

[1]

- (ii) State **one** similarity and **one** difference between Cas9 and the type of enzyme in (i).

similarity

difference

[2]

7 Which of the following statements about DNA replication in eukaryotes is/are correct?

- 1 Helicase exposes the templates required by DNA polymerase.
- 2 Free nucleotides are polymerised in the 3' to 5' direction.
- 3 Polymerisation of free nucleotides is continuous on both templates.

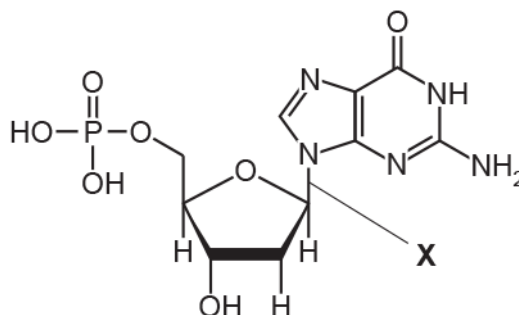
- A 1, 2 and 3 are correct
B only 1 and 2 are correct
C only 2 and 3 are correct
D only 1 is correct

Your answer

☐

[1]

8 The diagram below shows the structure of a nucleotide. A bond is labelled X.



Which of the following statements is/are correct?

- 1 X is a phosphodiester bond.
- 2 The sugar is deoxyribose.
- 3 The base is a purine.

- A 1, 2 and 3 are correct
B only 1 and 2 are correct
C only 2 and 3 are correct
D only 1 is correct

Your answer

☐

[1]

The article describes the use of flow cytometry ('automated blood analysers') to identify genetic variants linked to the risk of developing multiple myeloma.

Explain how flow cytometry could be used to identify genetic variants linked to the risk of developing multiple myeloma.

[4]

- Explain how a single gene mutation causes sickle cell anaemia.

[3]

- (c) The goal of the researchers at the International Myeloma Foundation is to identify people who develop multiple myeloma after having monoclonal gammopathy of undetermined significance (MGUS).

Evaluate the advantages and disadvantages of this approach compared with pedigree analysis.

[4]

(d) In MGUS, B cells produce an abnormal polypeptide called M protein.

Production of M protein involves the processes of transcription and translation.

Complete the table by putting a tick (✓) in the appropriate box to indicate whether the feature is involved in transcription, translation, both or neither.

Feature	Transcription only	Translation only	Both	Neither
C pairs with G				
A pairs with T				
Phosphodiester bonds are made				
Peptide bonds are made				

[4]

10 A student is purifying DNA from a bacterial culture.

Which of the molecules, A to D, is required for the precipitation of DNA during the purification process?

A ethanol

B glucose

C lysozyme

D water

Your answer

[1]

- (i) The roots of bean plants form nodules due to infection by the nitrogen-fixing bacteria, *Rhizobium*.

In response to infection by *Rhizobium*, bean plant nodule cells produce protein called leghaemoglobin.

Researchers wanted to find out more about three genes that code for leghaemoglobin.

They used RNA interference (RNAi) to inhibit the production of leghaemoglobin using miRNA. They measured the relative transcript level of the leghaemoglobin genes of bean plants treated with miRNA (RNAi plants) and those of untreated bean plants.

The results are shown in Table 1.

Name of leghaemoglobin gene	Relative transcript level of gene	
	Untreated plants	RNAi plants
LjLb1	3.5	0.085
LjLb2	4.0	
LjLb3	2.0	0.045

Table 1

Transcript levels for gene LjLb2 in the RNAi plants were reduced by 97.4% compared with the untreated plants.

Calculate the relative transcript level for LjLb2 in the RNAi plant.

Show your working.

Answer = ... [3]

12(a)

Fig. 4 shows a simplified diagram of part of a DNA molecule.

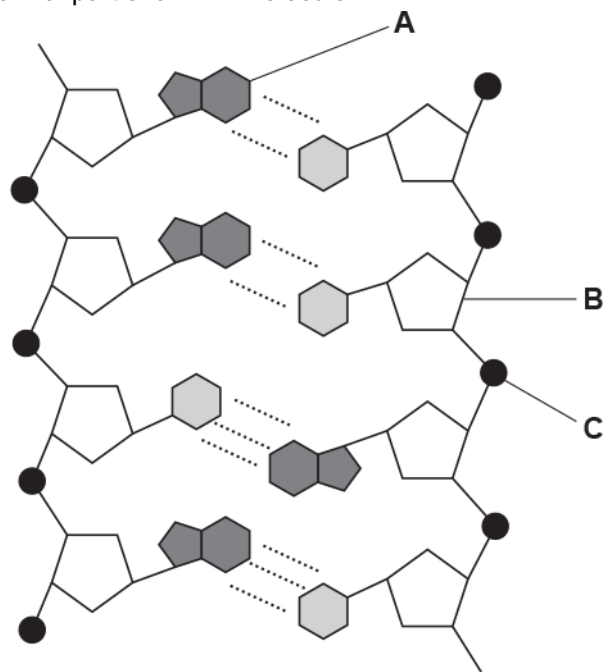


Fig. 4

(i) Identify the parts of the DNA molecule labelled A to C in Fig. 4.

A -----
B -----
C -----

[3]

(ii) The table below shows the results of an analysis of the base composition for each strand of a DNA molecule.

Complete the table by adding the missing values for strands 1 and 2.

DNA strand	Percentage of each base			
	A	C	G	T
strand 1		35	22	
strand 2	18			

[2]

(b) Hydrogen bonding is central to the function of nucleic acids.

Explain how hydrogen bonding contributes to the process of semi-conservative replication of DNA.

[2]

13 A gene mutation causes part of a DNA sequence to change from TAC to TAA.

TAA is a stop codon.

Which of the statements, **A** to **D**, is a correct explanation of why the stop codon may not affect translation of the mRNA sequence?

- A TAA also codes for an amino acid
- B TAA stops transcription, not translation
- C the mutation in the mRNA sequence may be repaired
- D the stop codon may not be present in the mature mRNA sequence

Your answer

[1]

14 Haemophilia A is a disease caused by a mutation in the gene coding for Factor VIII.

Factor VIII is involved in the blood clotting cascade.

(i) Scientists have found different types of mutation that can cause haemophilia A.

One type of mutation is a deletion of **two** bases. Suggest how this mutation causes the Factor VIII protein to lose its biological function.

----- [2]

(ii) Explain why haemophilia A is more common in males than in females.

----- [2]

(iii) A male with haemophilia A and a female who carries the disease have a child.

Complete the Punnett square below to determine the probability that the child will have haemophilia A. Use H/h to represent the alleles coding for Factor VIII.

		male	
			
female			
			

15 Adenosine triphosphate (ATP) is the principal source of energy in biological systems.

Fig. 32.1 is a diagram of the structure of an ATP molecule.

Two components of the molecule are labelled X and Y.

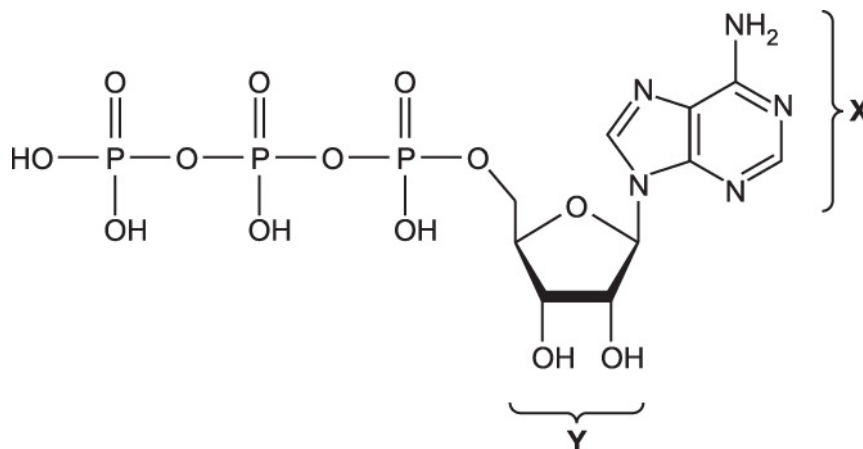


Fig. 32.1

(i) Name the parts of the molecule labelled X and Y in Fig. 32.1.

X

Y

[2]

(ii) With reference to the structure of ATP shown in Fig. 32.1, explain how this molecule provides chemical energy.

.....
.....
.....
.....
.....
.....

[3]

An experiment was conducted to investigate the roles of ATP and Ca^{2+} ions on the contraction of skeletal muscle.

The method was as follows:

1. Prepare four solutions, **A** to **D**
2. Cut frog skeletal muscle to a length of exactly 20 mm
3. Separate muscle into thin strands by cutting parallel to muscle fibres
4. Bathe muscle strands in glycerine (to make the cell membranes more permeable)
5. Place three muscle strands in solution **A** for 1 minute
6. After 1 minute, measure the length of each strand
7. Calculate the mean length of the muscle strands
8. Repeat steps 5-7 for solutions **B** to **D**.

Table 32.1 shows the chemical components that were present (✓) and absent (x) in solutions **A** to **D**.

Solution	ATP	Ca^{2+} ions	EDTA
A	x	✓	x
B	✓	x	x
C	✓	✓	x
D	✓	✓	✓

(iii)

Table 32.1

The results are shown in Fig. 32.2.

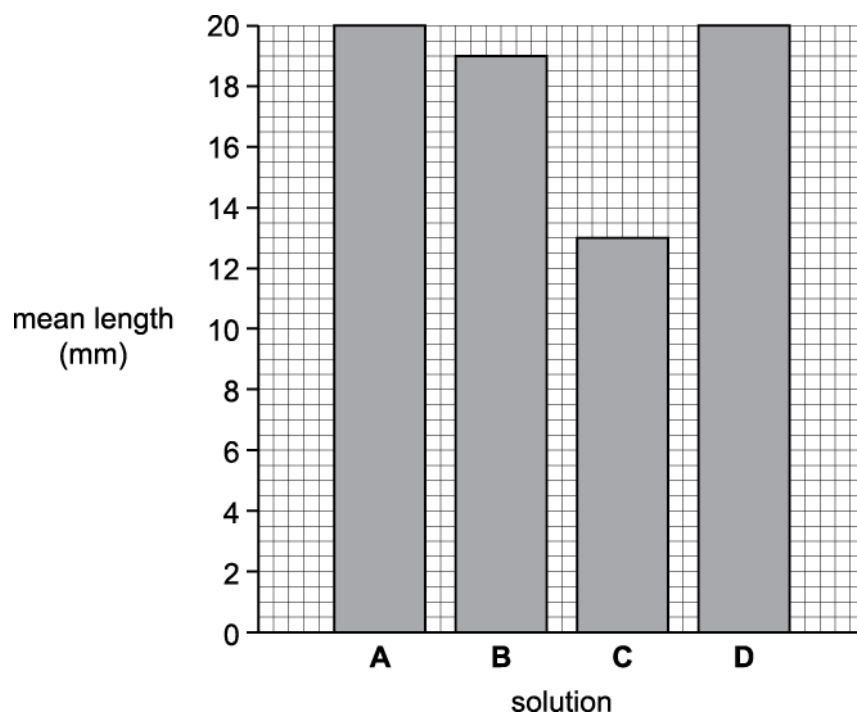
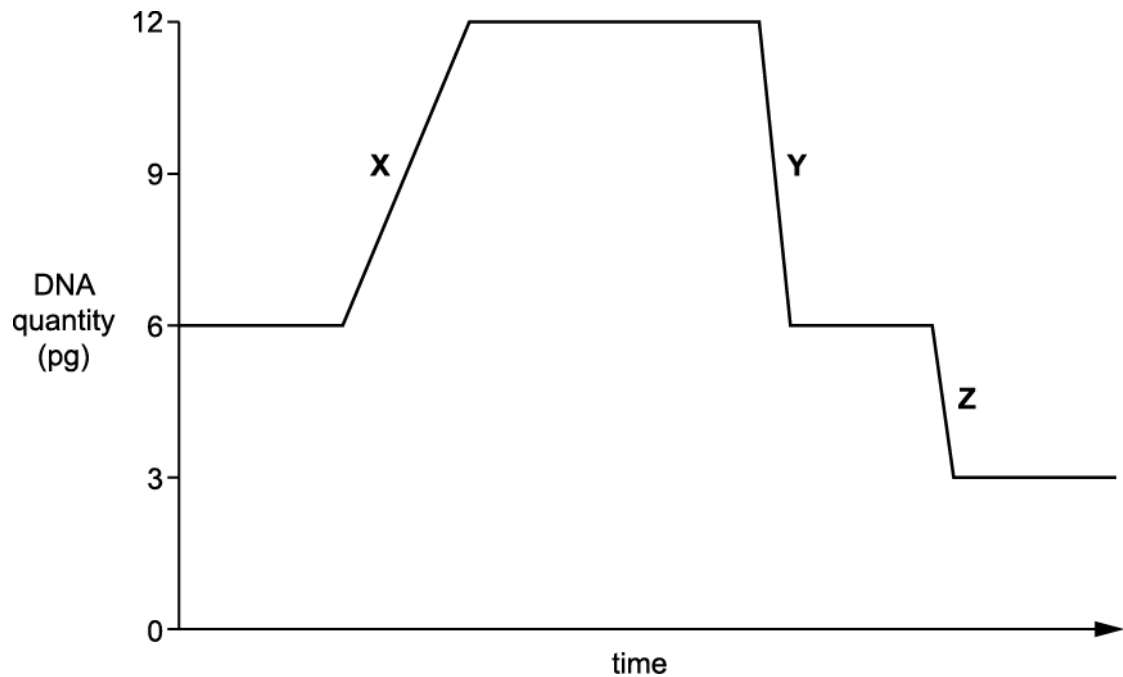


Fig. 32.2

- 16 The quantity of DNA in a cell changes throughout the cell cycle. The graph below shows these changes in a primary spermatocyte.

Three points on the graph are labelled X, Y and Z.



Which of the following statements is/are correct?

- 1 X represents the semi-conservative replication of DNA.
- 2 Z would result in the formation of a spermatid.
- 3 Y and Z represent telophase.

- A 1, 2 and 3 are correct
B Only 1 and 2 are correct
C Only 2 and 3 are correct
D Only 1 is correct

Your answer

[1]

17 A DNA sequence is composed of 30 percent guanine.

Which of the options, **A** to **D**, is the percentage of the sequence that is composed of adenine?

- A 20
- B 30
- C 60
- D 70

Your answer

[1]

18(a) This question is based on the Advance Notice article **CATALASE**.

Catalase is a homotetramer molecule found in human cells. Homotetramers are molecules that contains four identical subunits.

- (i) What evidence in the Advance Notice article confirms that human catalase must be a homotetramer? Explain your answer.

[2]

- (ii) Name one other tetrameric protein molecule found in human cells **and** state one similarity and one difference between the structure of this protein molecule and the structure of catalase.

protein -----

similarity -----

difference -----

[3]

- (b) Uric acid is an excretory product formed from the breakdown of purines.

- (i) Name **two** molecules other than DNA or RNA which contain a purine.

[2]

- (ii) Suggest why the removal of free radicals by uric acid might reduce the risk of cancer.

[1]

20(a) Choroideremia is an inherited sex-linked recessive condition that results in degeneration of the choroid layer in the eye. This condition leads to a gradual breakdown of the retina and eventual blindness.

Using the normal conventions for constructing genetic diagrams and the letters **E** and / or **e**, choose appropriate symbols to represent:

(i) the allele for choroideremia

-----[1]

(ii) the possible genotype(s) of a person who develops choroideremia

-----[1]

(iii) the genotype of a carrier of choroideremia.

-----[1]

(b) The gene involved in choroideremia codes for a protein called REP-1.

Many of the gene mutations that cause choroideremia result in the formation of a protein that is much smaller than normal REP-1. This smaller protein is known as a **truncated protein**.

(i) State the **organelle** in the cell where the following occurs:

a complementary RNA copy of the gene is synthesised -----

the REP-1 protein is synthesised -----

[2]

(ii) Suggest **how** a mutation in the REP-1 gene could lead to the formation of a truncated protein.

-----[2]

(c) A phase 1 clinical trial was carried out using gene therapy to treat choroideremia. An outline of the method used is given below.

- A complementary DNA (cDNA) copy of the REP–1 gene was inserted into a viral vector.
- The retina of the patient was detached to expose the choroid layer.
- A fine needle was used to inject the virus into the choroid layer.

(i) Name the enzyme used to create a cDNA copy of the REP–1 gene.

-----[1]

(ii) What type of gene therapy has been used in the trial?

-----[1]

(iii) Discuss the reasons why genetic diseases such as choroideremia are good choices for treatment using gene therapy.

-----[3]

21(a) This question is based on the Advanced Notice article **CHANGES IN THE NUCLEUS THAT LEAD TO CANCER**, which is an insert.

There are several different types of gene mutation. In substitution mutations one nucleotide is exchanged for another which produces a different base sequence.

Fig.1.1 represents part of a gene. The letters X and Y indicate two nucleotides that have been changed by a mutation.

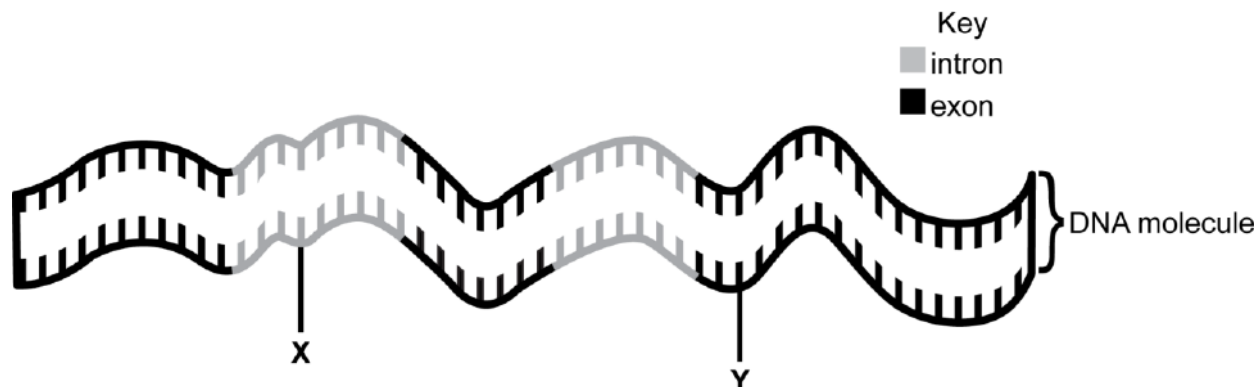


Fig. 1.1

(i) Name one genetic disease caused by a **substitution** mutation. [1]

(ii) Explain why a substitution mutation at point X will **not** result in a genetic disease. [2]

(iii) The substitution mutation at point Y on the DNA molecule resulted in a change from the triplet AGA to AGG. This did **not** result in a genetic disease.

Suggest why **this** substitution mutation did **not** result in a genetic disease. [2]

(b) Extract 2 of the Advanced Notice article describes the methylation of DNA.

Methyl groups are added to the cytosine in cytosine-guanine dinucleotides (CpGs) by the enzyme DNMT.

Fig.1.2 shows the structure of a section of DNA that has been methylated.

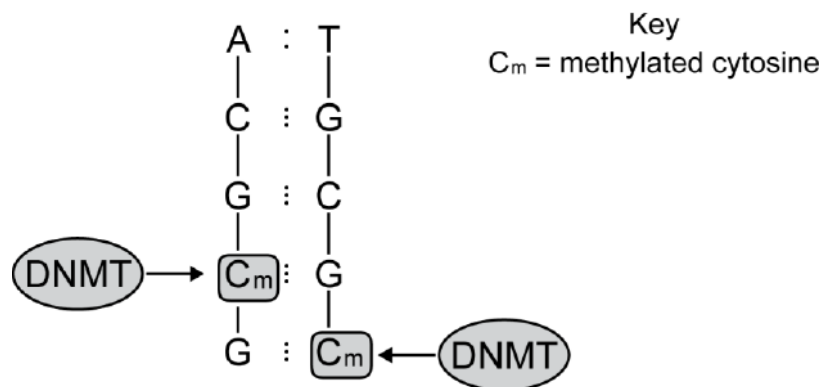


Fig. 1.2

(i) A nucleotide containing cytosine and a nucleotide containing guanine can be bonded together in two ways:

- in a dinucleotide
- in a base pair.

How are these bonds different?

[2]

(ii) Fig. 1.3 shows a sugar bonded to a molecule of cytosine which has been methylated by DNMT.

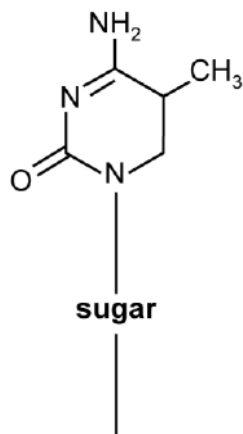


Fig. 1.3

Name the sugar to which the methylated cytosine is attached.

-----[1]

- (iii) The addition of a methyl group to cytosine increases the chances of the molecule being **deaminated** and converted to thymine.

On Fig.1.3, circle the part of the molecule that would be removed by deamination.

[1]

(c) In human DNA, the frequency of CpG sites across the genome varies.

In the human genome:

- analysis of the base content of human DNA shows that 21% of the bases present are cytosine.
- the **overall** frequency of CpG sites in the human genome is 0.01.
- in regions of the genome known as **promoters**, the frequency of CpGs is approximately 0.6.
- deamination of cytosine at CpG sites converts the cytosine to **thymine**.

(i) Using the information about the human genome, calculate the **expected** frequency of a CpG site in human DNA.

Show your working. Give your answer to **three** significant figures.

Answer = _____ [2]

(ii) By analysing the information about the human genome and your answer to (i), explain why the frequency of CpG sites is low in some regions but high in others.

[4]

- (d) In the Advanced Notice article you were told that HDAC and DNMT inhibitors are now available and used as anticancer drugs.

Fig.1.4 shows a section of DNA and its associated histone proteins.

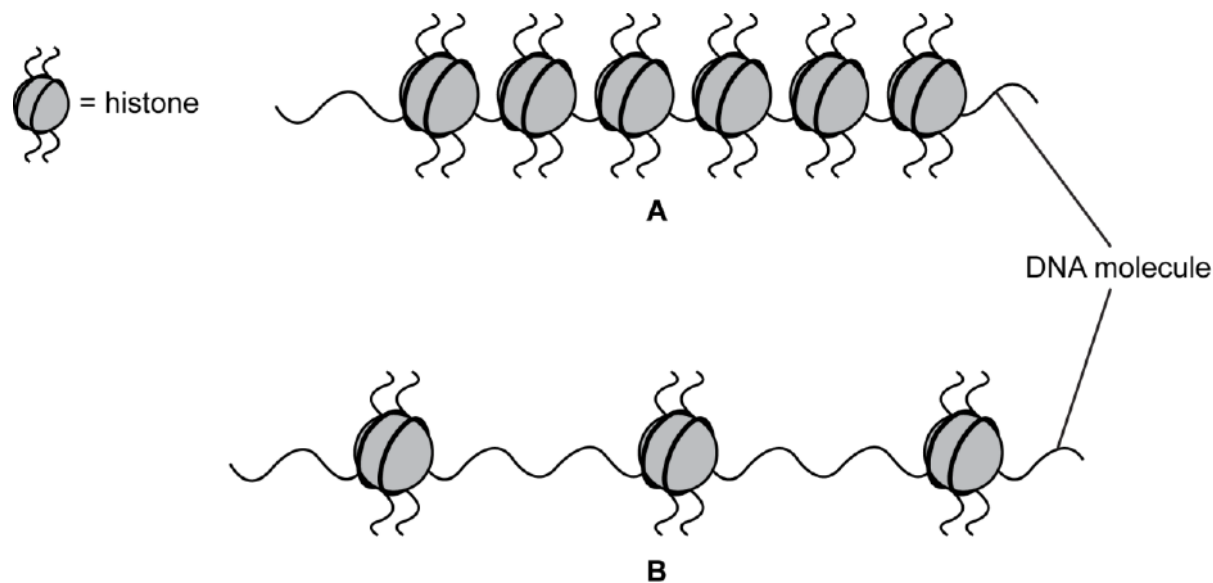


Fig. 1.4

- (i) Which of the two diagrams in Fig.1.4, **A** or **B**, shows the DNA and histones from a cancer cell that has been successfully treated with an HDAC inhibitor? Explain your answer.

[2]

- (ii) Inactivation of DNMT during DNA replication leads to the **loss** of methyl groups from DNA in dividing cancer cells.

Suggest why.

[1]

22 The GNA11 gene codes for a protein that is involved in cell signalling.

(i) Outline the changes in DNA which would lead to a faulty version of the GNA11 gene.

-----[1]

(ii) Suggest how the faulty version of the GNA11 gene is formed and suggest how the faulty GNA11 causes the development of ocular melanoma.

-----[3]

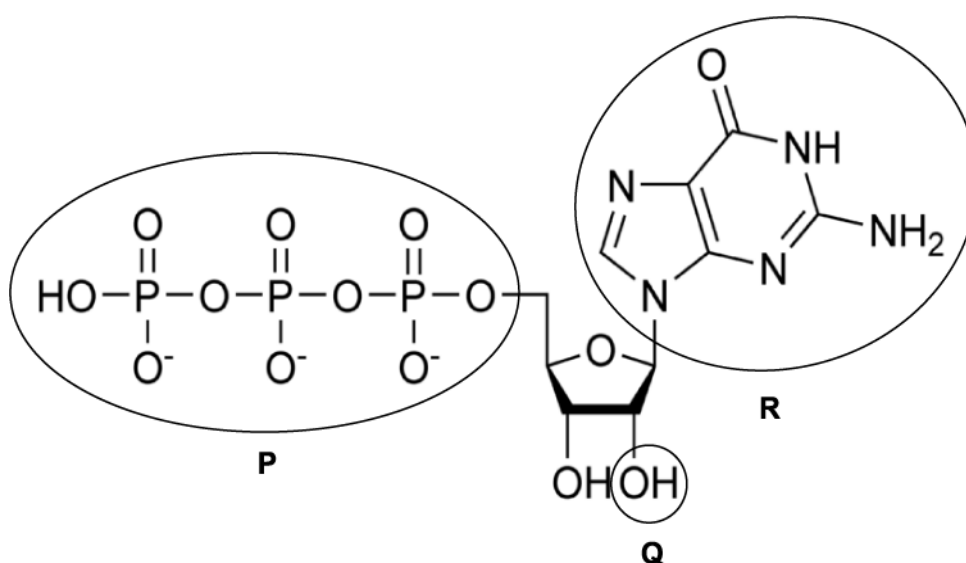
- 23 Thirty-three human blood group systems are known to exist. Two of these are the ABO blood group system and the Hh blood group system.

Explain why a person whose blood group is AB expresses both A and B antigens on the surface of their red blood cells.

----- [2]

- 24 The structure of a guanosine triphosphate (GTP) molecule is shown below.

Three regions of the molecule have been circled and labelled P, Q and R.



Which of the options, A to D, gives the regions that are structurally different to an adenosine monophosphate (AMP) molecule?

- A P, Q and R
- B P and Q
- C P and R
- D Q and R

Your answer

[1]

25 A length of template DNA has the following sequence: GATACTCCTAG.

Which of the options, **A** to **D**, is the mRNA sequence that would be transcribed from this length of DNA?

- A CTAGGAGTATC
- B CUAGGAGUAUC
- C CTATGAGGATC
- D CUAUGAGGAUC

Your answer

[1]

* Describe how a messenger RNA copy of a gene is produced.

* Describe how a messenger RNA copy of a gene is produced.

[6]

- (b) One mechanism for controlling gene expression in cells uses small, double stranded pieces of RNA known as siRNA.

- 1 siRNA molecules are introduced into the cell.
- 2 The siRNA molecules are combined with a protein complex called the RNA induced silencing complex (RISC) and one of the siRNA strands is destroyed.
- 3 The other strand remains bound to RISC and acts as a guide. RISC is now said to be activated.
- 4 This strand binds to complementary sequences on messenger RNA molecules in the cytoplasm causing them to be destroyed.

Fig. 36.1 shows the sequence of events for this mechanism.

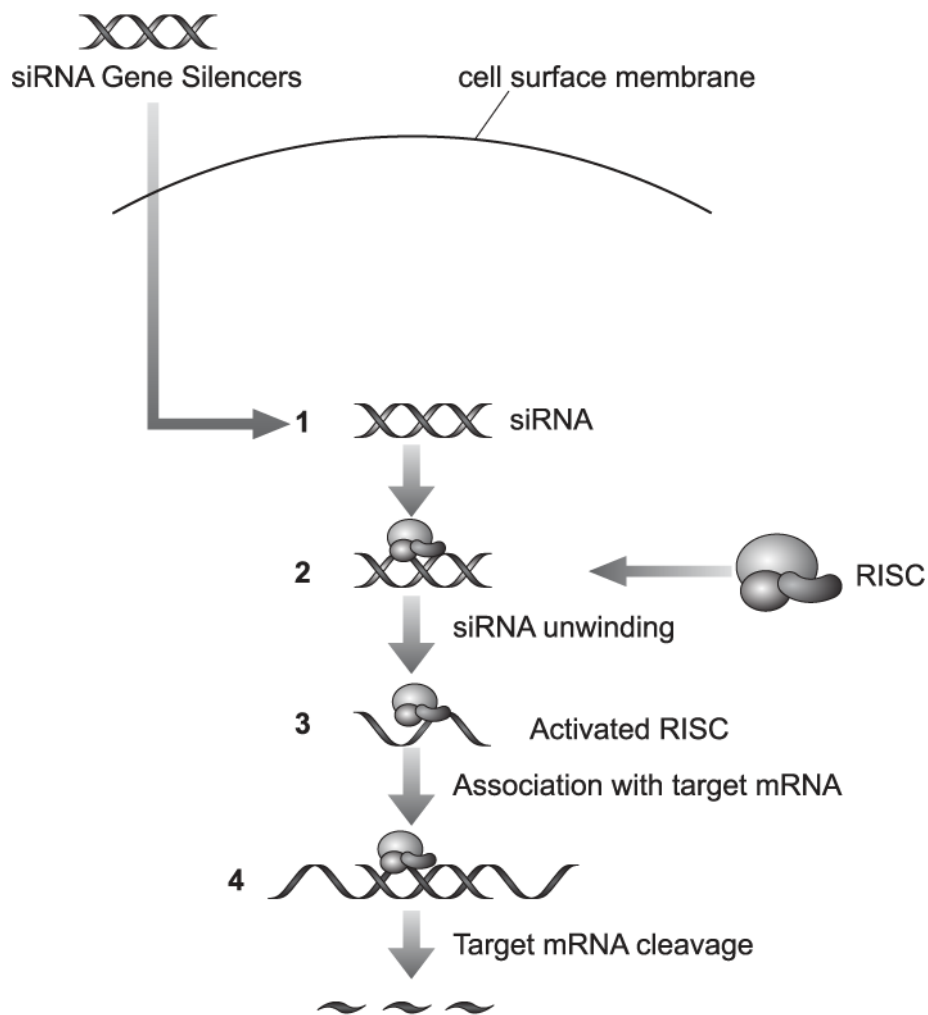


Fig. 36.1

- (i) An RNA strand on an activated RISC has the following sequence.

On the line above, write out the sequence that activated **RISC** would bind to on the mRNA molecule.

[1]

(ii) What type of reaction is carried out by activated **RISC** on the bonds in the mRNA molecule?

[1]

(iii) Clinical trialling is being carried out on the use of siRNA as an anti-viral therapy for the treatment of Hepatitis C infections.

Suggest how siRNA could prevent the spread of a virus within a person infected by the Hepatitis C virus.

----- [3]

27 Fig. 24.1 shows the structure of an ATP molecule.

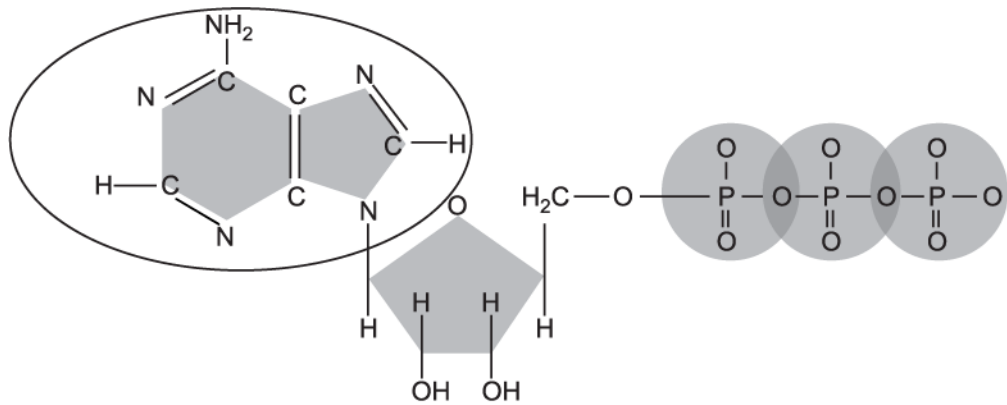


Fig. 24.1

Which part of the ATP molecule is circled?

- A adenine
- B adenosine
- C ribose
- D deoxyribose

Your answer ☐

[1]

END OF QUESTION PAPER

Mark Scheme

Question			Answer/Indicative content	Marks	Guidance
1	a	i	carries, genetic information / code , from nucleus to ribosomes ✓	1(AO1.1)	ALLOW a correct description of mRNA in a mitochondria or prokaryotic cell
		ii	carries specific amino acid / forms complex with amino acid AND anticodon binds to codon on mRNA ✓	1(AO1.1)	ALLOW descriptions of anticodon and codon
	b		genes ✓ three ✓ degenerate ✓ overlap ✓	4(AO1.1)	
			Total	6	
2		i	DNA has phosphate (groups) in the backbone ✓ phosphate (group) is charged ✓ <i>idea that</i> forces of attraction occurs between opposite charges on phosphate and arginine ✓	2 max(AO2.1)	ALLOW has sugar-phosphate backbone
		ii	enables sperm nucleus to be small ✓ greater protection of DNA molecules ✓	1 max(AO2.1)	ALLOW enables the head of the sperm to be small
			Total	3	
3	a	i	any two from: (mtDNA is) circular ✓ (mtDNA is) not associated with , histones / proteins ✓ (mtDNA does) not contain (much / many) , introns / junk DNA ✓	2 AO2.1	ALLOW any of the differences described in the article, except for size / number of base pairs (this is excluded by the question stem)

Mark Scheme

Question			Answer/Indicative content	Marks	Guidance																				
		ii	<table><tr><td>Component</td><td>mtDNA</td><td>RNA</td><td>ATP</td></tr><tr><td>adenine</td><td>✓</td><td>✓</td><td>✓</td></tr><tr><td>ribose</td><td></td><td>✓</td><td>✓</td></tr><tr><td>uracil</td><td></td><td>✓</td><td></td></tr><tr><td>pyrimidine</td><td>✓</td><td>✓</td><td></td></tr></table>	Component	mtDNA	RNA	ATP	adenine	✓	✓	✓	ribose		✓	✓	uracil		✓		pyrimidine	✓	✓		3 AO1.1 2.1	1 mark per correct row
Component	mtDNA	RNA	ATP																						
adenine	✓	✓	✓																						
ribose		✓	✓																						
uracil		✓																							
pyrimidine	✓	✓																							
		iii	(mitochondrial) <u>tRNA</u> / <u>rRNA</u> ✓	1 AO2.1	DO NOT ALLOW ‘transcription factors’ unless it is made clear that these are RNA transcription factors. IGNORE ‘transport’																				
	b	i	(production of) two identical DNA molecules ✓ using each strand as a template / where half the genetic information is kept in the new DNA ✓	2 AO1.1																					
		ii	mutation in catalytic region , prevents / AW, mtDNA replication ✓ mutation in exonuclease (domain) increases mutation rate ✓	2 AO2.1	ALLOW reduces accuracy of replication or causes damaged DNA to be replicated DO NOT ALLOW just mismatched base-pairs may not be removed																				
			Total	10																					
4		i	hydrogen ✓	1 AO1.2																					
		ii	phosphate (group) AND <u>deoxyribose</u> ✓	1 AO1.2	Both required for 1 mark.																				
			Total	2																					

Mark Scheme

Question			Answer/Indicative content	Marks	Guidance
5			D	1 AO2.4	
			Total	1	

Mark Scheme

Question			Answer/Indicative content	Marks	Guidance
6	a		same DNA sequence in opposite (5' to 3') direction on both strands ✓	max 1 AO1.1	DO NOT ALLOW sequence that reads the same in 5' to 3' and 3' to 5' directions <u>Examiner's Comments</u> Only a few candidates were able to answer this question, as it proved to be challenging. Many simply stated that it is a sequence of nucleotide bases that reads the same forwards as backwards. In the context of DNA however candidates need to refer to the two strands of DNA and be clear that the bases are in opposite direction on each strand. Sometimes a diagram was useful in this answer.
	b	i	restriction (enzyme / endonuclease) ✓	1 AO1.1	
		ii	<i>similarity</i> cut at, <u>precise / specific</u>, sites / DNA sequences ✓ cut both strands of DNA ✓ both hydrolyse phosphodiester bonds ✓ <i>difference</i> Cas9, is guided / requires gRNA, but restriction enzymes function alone ✓ Cas9, cuts straight / produces blunt ends, but some restriction enzymes produce sticky ends ✓	max 2 AO2.1	must have similarity and difference for 2 marks <u>Examiner's Comments</u> A lot of candidates didn't score any marks in this question. This was mainly because they missed out the statement in the question to (b)(i) which stated that all endonucleases cut within a nucleotide sequence. Candidates should have used this to help them answer (b)(ii). Exemplar 4 similarity <u>they both cut DNA strands, inactivating a gene</u> difference <u>restriction enzymes produce sticky ends via a staggered cut.</u> [2] In Exemplar 4 the first point did not give enough detail about the nature of the cut and the differences were not credited because the candidate did not compare both enzymes and only described what the restriction enzyme did.
			Total	4	

Mark Scheme

Question			Answer/Indicative content	Marks	Guidance
7			D	1 AO1.2	
			Total	1	
8			C	1 AO2.1	
			Total	1	

Mark Scheme

Question			Answer/Indicative content	Marks	Guidance
9	a		Label cells with fluorescent marker ✓ (fluorescence attached to) antibodies ✓ (antibodies bind to) <u>antigens</u> on (white blood) cells ✓ (alternative) fluorescent DNA probe ✓ counts / sorts, cells (with genetic variant) ✓	4 max AO2.3	IGNORE red blood cells IGNORE fluorescent marker binds to antigens ALLOW section of DNA for 'DNA probe' <u>Examiner's Comments</u> Candidates clearly recognised this technique and most candidates discussed the counting of cells and the use of fluorescence. To achieve full marks, candidates had to follow a sequential logic of the labelling technique. Some candidates did refer to DNA probes as they made the connection between searching for genetic variants and the direct use of these probes.

Mark Scheme

Question		Answer/Indicative content	Marks	Guidance
	b	base / nucleotide, substitution ✓ mRNA codon changes OR different amino acid (attached) ✓ changes haemoglobin, (tertiary) structure / (3D) shape ✓ (abnormal) haemoglobin, clumps / crystallises (clumping) at low O₂ concentration ✓ (only present in) homozygous recessive individuals ✓	3 max AO1.1	ALLOW named bases ALLOW glutamic acid changed to valine ALLOW different primary structure IGNORE mutations in haemoglobin protein IGNORE red blood cells clump together ALLOW H^S H^S for 'homozygous recessive' <u>Examiner's Comments</u> Many candidates clearly knew the amino acid substitution causing sickle cell anaemia but often struggled to describe the type of mutation. Many candidates recognised it as a substitution but did not state base/nucleotide thus not differentiating it from the mark for amino acid substitution. Candidates then struggled to relate this change to an alteration in haemoglobin, referring straight to the altered erythrocyte shape. A few candidates discussed the gene mutation with the erythrocyte directly, confusing a cell with the protein inside.  OCR support OCR delivery guide on Population genetics and epigenetics (5.1.2) can offer support: https://www.ocr.org.uk/qualifications/as-a-level-gce-biology-b-advancing-biology-h022-h422-from-2015/delivery-guide/module-bb05-module-5-genetics-control-and-homeostasis/delivery-guide-bbdg028-population-genetics-and-epigenetics-512

Mark Scheme

Question			Answer/Indicative content	Marks	Guidance																									
	c		<i>advantages of the IMF approach:</i> (may) give a more accurate diagnosis ✓ treat / intervene, earlier ✓ identify people (at risk) who do not have family history OR removes worry for people that do have a family history ✓ <i>disadvantages of IMF approach:</i> results of the MGUS screening may give false positives ✓ more expensive (than pedigree analysis) ✓	4 max AO3.2	ALLOW ora throughout ALLOW prevents / cures, myeloma <u>Examiner’s Comments</u> Candidates knew the relevance of pedigree analysis and answered this question from this perspective. It was clear the pre-release material had been read as the most common mark point was the expense of this technique from this section of the pre-release; ‘<i>Researchers at the IMF store thousands of blood These expensive tests allow the identification of genetic variants that might be linked to the risk of developing multiple myeloma.</i>’ Many candidates also recognised the relevance of the ‘identification’ aspect of this quote and stated this approach was more accurate. All mark points were seen as some candidates not only used the pre-release material to gain information but realised the limitations of a pedigree analysis to those who have a family history and would not detect those who did not have this history.																									
	d		<table><tr><th>Feature</th><th>Transcription only</th><th>Translation only</th><th>Both</th><th>Neither</th></tr><tr><td>C pairs with G</td><td></td><td></td><td>✓</td><td></td></tr><tr><td>A pairs with T</td><td>✓</td><td></td><td></td><td></td></tr><tr><td>Phosphodiester bonds are made</td><td>✓</td><td></td><td></td><td></td></tr><tr><td>Peptide bonds are made</td><td></td><td>✓</td><td></td><td></td></tr></table>	Feature	Transcription only	Translation only	Both	Neither	C pairs with G			✓		A pairs with T	✓				Phosphodiester bonds are made	✓				Peptide bonds are made		✓			4 AO2.1	Mark the first answer. If an additional answer is given that is incorrect, then = 0 marks ALLOW crosses or any other unequivocal symbol as alternative to ticks. <u>Examiner’s Comments</u> Few candidates achieved full marks, with many candidates scoring at least 2 marks. Most candidates did not realise that the T base would not be present during translation and that the only covalent bonds made during translation are peptide bonds.
Feature	Transcription only	Translation only	Both	Neither																										
C pairs with G			✓																											
A pairs with T	✓																													
Phosphodiester bonds are made	✓																													
Peptide bonds are made		✓																												
			Total	0																										
10			A	1																										
			Total	1																										

Mark Scheme

Question			Answer/Indicative content	Marks	Guidance
11		i	0.104 ✓✓✓	3	If answer is incorrect or missing, a maximum of 2 marks can be given for intermediate stages as follows: one mark for working such as • $4.0 - ((97.4 / 100) \times 4.0)$ • 4 & 0.026 • $100 - 97.4 = 2.6\%$ with 2.6% of 4.0 = 0.104 one mark for incomplete calculation • 3.896 one mark for correct answer but not quoted to 3dp e.g. 0.10 <u>Examiner's Comments</u> Candidates should provide answers that are to the same number of decimal places for other data in the same column. In this case the answer should be given to 3 decimal places.

Mark Scheme

Question	Answer/Indicative content	Marks	Guidance
ii	inhibits <u>translation</u> of mRNA ✓ (miRNA) binds at a <u>complementary</u> site (on mRNA) ✓ argonaute protein, breaks/cleaves, the mRNA strand ✓ AVP ✓	3 max	IGNORE references to 'miRNA inhibits mRNA' as this is given in the stem of the question DO NOT ALLOW references to inhibiting transcription Further detail e.g. • double stranded precursor binds to, dicer / endonuclease protein • dicer cuts precursor (into short segments) • dicer cuts precursor • (short double stranded) miRNA binds to argonaute protein • RNA induced silencing complex (RISC) formed • small sections of mRNA can be translated but will not result in formation of a, functional / complete, protein Examiner's Comments Some candidates did not score marks in this questions due to poor expression; for exampling muddling mRNA and miRNA in their answers. Others gave answers that related to enzyme inhibition which did not relate to the question. This area of the specification is new and examiners observed many answers that were lacking depth and detail. Exemplar 2 Double stranded RNA binds to a dicer which cleaves the RNA to around 200 nucleotide lengths. An argonaute protein binds to one of the RNA strands - RNA strand has a complementary sequence of nucleotides which will have complementary base pairing to the target mRNA. It binds from between mRNA and mRNA, miRNA cleaves the mRNA and prevents it from being translated.

Mark Scheme

Question			Answer/Indicative content	Marks	Guidance
		iii	miRNA less, precise / specific (than siRNA) ✓	1	DO NOT ALLOW 'miRNA is more accurate' ALLOW miRNA will bind to more than one mRNA ALLOW miRNA will inhibit, all / more, of the leghaemoglobin genes Examiner's Comments Again some less able candidates discussed enzyme inhibition rather than the difference between precision of miRNA and siRNA. Exemplar 3 <i>miRNA is more specific acting - that it will only inhibit one specific gene. siRNA affects is able to affect siRNA is more specific. miRNA is able to inhibit several of the genes at once / siRNA may only inhibit one.</i>
			Total	7	

Mark Scheme

Question			Answer/Indicative content	Marks	Guidance																			
12	a	i	A adenine / <u>purine</u> ✓ B <u>deoxyribose</u> / pentose ✓ C phosphate ✓	3	Examiner's Comments (a)(i) was well answered but with a few candidates stating nitrogenous base only for A.																			
		ii	<table><tr><th rowspan="2">DNA strand</th><th colspan="4">Percentage of each base</th></tr><tr><th>A</th><th>C</th><th>G</th><th>T</th></tr><tr><td>strand 1</td><td>25</td><td>(35)</td><td>(22)</td><td>18</td></tr><tr><td>strand 2</td><td>(18)</td><td>22</td><td>35</td><td>25</td></tr></table> ✓✓	DNA strand	Percentage of each base				A	C	G	T	strand 1	25	(35)	(22)	18	strand 2	(18)	22	35	25	2	2 rows correct = 2 marks 1 row correct = 1 mark Examiner's Comments Many candidates achieved full marks for (a)(ii). Errors appeared to be random and showed some candidates did not read the heading of percentage and that each row had to add up to 100.
DNA strand	Percentage of each base																							
	A	C	G	T																				
strand 1	25	(35)	(22)	18																				
strand 2	(18)	22	35	25																				
	b		hydrogen bonds broken by <u>helicase</u> (to separate the strands) ✓ hydrogen bonds between free nucleotide and exposed (template) base ✓ 2 bonds between A & T and 3 between C & G ✓ complementary, bases / base pairing ✓ joins, DNA strands / polynucleotides / old strand and new strand ✓	2 max	DO NOT ALLOW hydrolyses hydrogen bond Examiner's Comments (b) was well answered with most candidates appreciating that complementary base pairing referred to the hydrogen bonding between bases. Some answers discussed DNA replication without focusing on the role of hydrogen bonding.																			
			Total	7																				
13			D ✓	1	Examiner's Comments This question was assessing whether candidates understood that not all the DNA sequence is transcribed into mature mRNA. The most common incorrect response was that repair of the mRNA took place.																			
			Total	1																				

Mark Scheme

Question			Answer/Indicative content	Marks	Guidance						
14		i	interrupts triplet code / causes frame-shift / production of premature stop codon ✓ (DNA) triplets code for different amino acids ✓ primary structure of protein changed ✓ protein cannot interact with other, proteins / named component ✓	max 2							
		ii	gene (encoding Factor VIII), on X chromosome / X-linked / sex-linked ✓ males lack second X chromosome ✓	2	ORA ORA						
		iii	male gametes female gametes <table><tr><td>X^H</td><td>$X^H X^h$</td><td>$X^H Y$</td></tr><tr><td>X^h</td><td>$X^h X^h$</td><td>$X^h Y$</td></tr></table> probability = 0.5 ✓✓	X^H	$X^H X^h$	$X^H Y$	X^h	$X^h X^h$	$X^h Y$	3	ALLOW H/h not superscripted on X ALLOW 50% or ½
X^H	$X^H X^h$	$X^H Y$									
X^h	$X^h X^h$	$X^h Y$									
			Total	7							
15		i	X adenine (base) ✓ Y ribose (sugar) ✓	2	DO NOT ALLOW adenosine						
		ii	transfer / release, of, <u>last / terminal</u> , phosphate (group) ✓ to produce, ADP / adenosine diphosphate ✓ via hydrolysis reaction ✓ breaking of high-energy bond between phosphates ✓	max 3	ALLOW release of, inorganic phosphate / Pi ALLOW ‘using a water molecule’						
			Total	5							
16			B ✓	1							
			Total	1							
17			A ✓	1							
			Total	1							

Mark Scheme

Question			Answer/Indicative content	Marks	Guidance
18	a	i	<i>evidence</i> - only one gene (locus) (paragraph 8) ✓ <i>idea that</i> one gene codes for one polypeptide ✓	2	
		ii	<i>protein</i> - haemoglobin ✓ <i>similarity</i> - <i>idea of</i> same prosthetic group / haem attached to each sub-unit ✓ <i>difference</i> - <i>idea that</i> there are two different sub-units in haemoglobin / 2 alpha and 2 beta sub-units ✓	3	IGNORE reference to same number of sub-units ALLOW 'haemoglobin is not a homotetramer' or 'haemoglobin is a heterodimer' IGNORE 'haemoglobin is a heterotetramer'
	b	i	ATP / ADP / AMP / NAD / NADH / FAD / FADH ✓✓	2	
		ii	<i>idea that free radicals</i> cause mutations within proto-oncogenes and/or tumour suppressor genes ✓	1	ALLOW DNA damage rather than mutation but the link to relevant genes must be present
			Total	8	

Mark Scheme

Question			Answer/Indicative content	Marks	Guidance
19		i	1 <i>idea that</i> (causes) a change in, a DNA, triplet / code / sequence; DNA triplet code / 2 (leading to) a change in, mRNA / codon; 3 (leading to) a change in, tRNA / anticodon (at ribosome); 4 (leading to) a different, amino acid / primary structure / sequence of amino acids; 5 (different amino acids have) different R groups; 6 different bond(ing) in (protein) tertiary structure; 7 <i>idea that</i> (change) leads to (protein) changing its shape / described; 8 consequence for protein action described;	5	ACCEPT idea of 'wrong' or 'incorrect' for 'different' or 'changed' throughout 1. ACCEPT mutation might lead to a stop codon inserted 2. CREDIT idea of a triplet or 3 mRNA bases 6. ACCEPT 'bonding / folding altered in tertiary structure' DO NOT CREDIT 'no protein made' 8 CREDIT examples e.g. (enzyme) no enzyme substrate complex forms, (antibody) variable region not binding to antigen, (protein channel) no hydrophilic R groups lining channel e.g. mutation might lead to a stop codon inserted
		ii	DMD is not a chromosome mutation; (karyotypes) (amniocentesis or CVS) detect, chromosome mutations / described;	1	IGNORE 'DMD is a gene mutation' (as this is given in the stem of the question) CREDIT description e.g. changes in chromosome number or structure.
			Total	6	

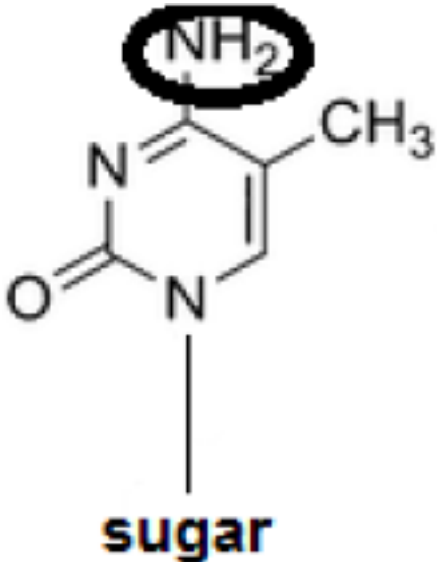
Mark Scheme

Question			Answer/Indicative content	Marks	Guidance
20	a	i	e;	1	CREDIT X ^e ACCEPT a single, lower case alternative letter IGNORE 'c' unless it is clearly 'lower case' IGNORE I ^e
		ii	X ^e Y and X ^e X ^e	1	ecf for the allele symbol in (i)
		iii	X ^E X ^e	1	ecf for the allele symbol in (i) Examiner's Comments While many were aware that 'e' would be the allele symbol for the condition, some put a genotype (ee) in (i) and only the better candidates went on to give the correct genotypes for (ii) and (iii).
	b	i	1. nucleus / nucleolus; 2. ribosomes;	1	ANSWERS MUST BE IN THIS ORDER ACCEPT RER / rough endoplasmic reticulum
		ii	<i>idea that</i> mutation (in DNA) leads to a, stop / termination, codon OR <i>idea that</i> DNA sequence is deleted OR <i>idea that</i> mutation leads to more RNA being spliced out; <i>idea that</i> translation is terminated by stop codon; no (more) amino acids are added (after the stop codon) / fewer amino acids (in truncated protein);	2 max	IGNORE ref to a base deletion in context of shorter DNA Examiner's Comments Part (i) was synoptic and answered well but in (ii) too many answers were given in terms of changing the sequence of amino acids rather than producing a protein with fewer amino acids.
	c	i	reverse transcriptase;	1	
		ii	somatic (gene therapy);	1	CREDIT augmentation (gene therapy)

Mark Scheme

Question			Answer/Indicative content	Marks	Guidance
		iii	<i>Idea that</i> disease involves a defect in a single gene; no other cause; <i>idea that</i> disease is easily identified in sufferers; gene / gene product, is known; gene can be delivered reliably to affected cells;	3	Mp2 ACCEPT idea that disease will be cured / vision will be restored. Mp3 CREDIT reverse argument 'easy to see if it has worked' ACCEPT 'easy to insert the gene' Examiner's Comments This was intended to be accessible so it was disappointing how many candidates did not 'spot' reverse transcriptase or the fact that this would be somatic gene therapy. In part (iii) many candidates did not pick up on the question rubric and answered in terms of germ line therapy or ethics. It would be a worthwhile discussion for Centres to have with their candidates as to what makes some genetic diseases targets for treatment using gene therapy while others are not.
			Total	12	

Mark Scheme

Question			Answer/Indicative content	Marks	Guidance
21	a	i	Sickle cell anaemia / PKU / Cystic Fibrosis	1	DO NOT ALLOW Huntington's ALLOW alternative correct suggestions
		ii	(X in intron) <i>idea that</i> code in this region does not appear in mature mRNA (1) <i>idea that</i> introns are transcribed but edited out (1)	2	ALLOW an explanation that this region does not code for amino acids
		iii	<i>idea that</i> genetic code is degenerate (1) <i>idea that</i> new triplet codes for an amino acid with a similar R group (1) no change in the tertiary structure of the protein (1)	2	ALLOW a description - some amino acids have more than one triplet ALLOW idea of 'little or no change' if second mark point is awarded ALLOW three dimensional protein structure DO NOT ALLOW 'no change in protein structure' without further qualification since 'structure' is given in the stem of the question
	b	i	(dinucleotide bond is) phosphodiester bond / covalent bond / formed by a condensation reaction (1) (base pair is) (3) hydrogen bonds (1)	2	
		ii	deoxyribose (1)	1	DO NOT ALLOW 'pentose' or 'ribose' as they are told that DNMT methylates DNA
		iii		1	

Mark Scheme

Question			Answer/Indicative content	Marks	Guidance
	c	i	0.044 = 2 marks (1)(1)	2	If answer is incorrect look for: 0.21 x 0.21 OR (21/100) x (21/100) OR 4.4% for max 1 mark
		ii	<i>Idea that overall frequency is much lower than expected (1)</i> <i>reason for 0.01 / low frequency</i> more risk of gene mutations occurring (1) <i>idea of base substitution (1)</i> <i>reason for 0.6 / high frequency</i> <i>idea that</i> promotor requires more methylation sites than other regions (1) <i>idea that</i> promoters regulate gene expression (1) <i>idea that</i> RNA polymerase binds to promoter when it is unmethylated (1) methylation allows genes to be switched off when not required (1)	4	DO NOT ALLOW the reverse argument as this is given in the case study
	d	i	<i>Fig. 1.4B (no mark)</i> <i>Explanation:</i> <i>idea that</i> histones still have acetyl groups present (1) <i>idea that</i> chromatin is more open / DNA is more accessible (1) <i>idea that</i> DNA is available for transcription (1) <i>idea that</i> (named) tumour suppressor gene is expressed (1)	2	Maximum of one mark if Fig. 1.4A given as answer ALLOW reverse argument 'acetyl groups have not been removed' ALLOW reference to expression of <i>p53</i> or <i>p16</i> or any correct tumour suppressor gene
		ii	<i>idea that</i> DNMT not available to methylate new strands	1	
			Total	18	

Mark Scheme

Question			Answer/Indicative content	Marks	Guidance
22		i	<i>idea of</i> changes in, base / nucleotide / triplet, sequence	1	
		ii	mutation / AW, to proto-oncogene (leads to) faulty, receptor / growth factor (leads to) uncontrolled cell division	3	ALLOW forms oncogene DO NOT ALLOW (cell) signalling molecule because this is referenced in the stem
			Total	4	
23			Any 2 from: (A and B are) codominant <i>idea that</i> both genes are transcribed <i>idea that</i> both antigen proteins are produced	2	
			Total	2	
24			C	1	
			Total	1	
25			D	1	
			Total	1	

Mark Scheme

Question			Answer/Indicative content	Marks	Guidance										
26	a		* Level 3 (5–6 marks) A detailed description, including at least one statement from each section in the correct sequence (from 1, 2 and then 3), showing knowledge and understanding of the production of mRNA is given and all steps are in the correct order. <i>There is a well-developed line of reasoning which is clear and logically structured. The information presented is relevant and substantiated.</i> Level 2 (3–4 marks) Description includes at least two correct statements in the correct sequence but there is some repetition or irrelevant information (e.g. about translation). <i>There is a line of reasoning presented with some structure. The information presented is in the most-part relevant and supported by some evidence.</i> Level 1 (1–2 marks) One correct statement is made which include points from any of the sections shown in the guidance. <i>The information is basic and communicated in an unstructured way. The information is supported by limited evidence and the relationship to the evidence may not be clear.</i> 0 marks No response or no response worthy of credit.	6	Sections indicate possible scientific points and the expected sequence of the answer. Section 1 - RNA polymerase binds to the DNA - reference to the promoter region (for binding) - reference to transcription factors OR DNA unwinding (by DNA helicase) Section 2 - reference to RNA, nucleotides / bases, pairing with bases on, template strand - reference to complementary base pairing / numbers of hydrogen bonds forming - correct reference to base pairings <table><tr><td>DNA</td><td>RNA</td></tr><tr><td>A</td><td>U</td></tr><tr><td>T</td><td>A</td></tr><tr><td>C</td><td>G</td></tr><tr><td>G</td><td>C</td></tr></table> - reference to RNA polymerase, catalysing / AW, formation of, phosphodiester bonds / AW Section 3 - reference to termination / stop sequence - reference to DNA rewinding / RNA polymerase leaving the DNA strand - reference to removal of introns from primary RNA / formation of, mature / messenger RNA	DNA	RNA	A	U	T	A	C	G	G	C
DNA	RNA														
A	U														
T	A														
C	G														
G	C														
	b	i	ACGGGAAGGGCCCGAGCACGGA UGCCCUUCCCGGGCUCGUGCCU	1											
		ii	hydrolysis	1	IGNORE 'cleavage'										

Mark Scheme

Question			Answer/Indicative content	Marks	Guidance
		iii	Any 3 from: (activated) RISC, cleaves / AW, viral mRNA no viral proteins made no, viral particles / AW, assembled <i>idea that</i> no new cells are infected	3	IGNORE 'virus cannot spread' as this is given in the question
			Total	11	
27			A	1	
			Total	1	