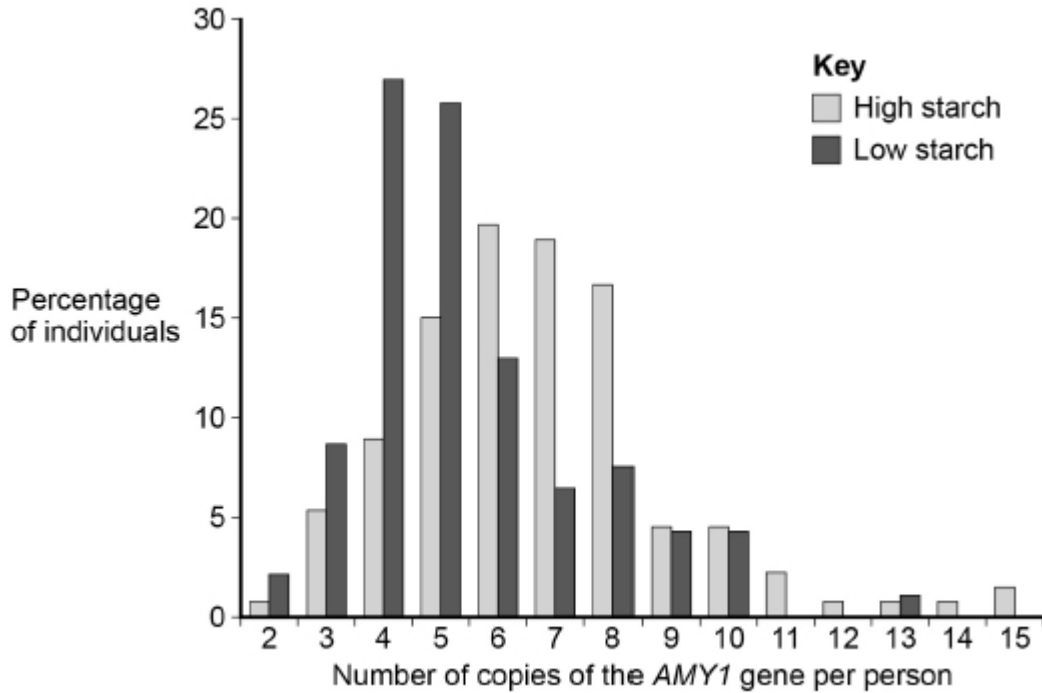


4.2 GENETIC INFORMATION, VARIATION AND RELATIONSHIP – DNA AND PROTEIN SYNTHESIS (1) – QUESTIONS

Q1. (a) Most human cells contain two copies of each gene. However, there might be up to 15 copies of the gene for amylase (*AMY1*). Scientists investigated the number of copies of the *AMY1* gene in individual people in two populations. One population had a high-starch diet and the other population had a low-starch diet.

The graph below shows their results.



Describe what their results show.

(3)

(b) Multiple copies of the *AMY1* gene is an adaptation to a high-starch diet.

Use your knowledge of protein synthesis and enzyme action to explain the advantage of this adaptation.

(3)

(c) Multiple copies of the *AMY1* gene is an adaptation to a high-starch diet.
Suggest how this evolved through natural selection.

(3)

(Total 9 marks)

Q2. (a) DNA is a polymer of nucleotides. Each nucleotide contains an organic base.

Explain how the organic bases help to stabilise the structure of DNA.

(2)

(b) Triplets of bases in a DNA molecule code for the sequence of amino acids in a polypeptide. The genetic code is frequently written as the three bases on mRNA that are complementary to a triplet on DNA. **Table 1** shows what different combinations of bases

on mRNA code for. The names of amino acids are abbreviated. For example, 'Ala' stands for alanine.

Table 1

First base	Second base								Third base
	Guanine (G)		Adenine (A)		Cytosine (C)		Uracil (U)		
G	GGG	Ala	GAG	Glu	GCG	Ala	GUG	Val	G
	GGA	Gly	GAA	Glu	GCA	Ala	GUA	Val	A
	GGC	Gly	GAC	Asp	GCC	Ala	GUC	Val	C
	GGU	Gly	GAU	Asp	GCU	Ala	GUU	Val	U
A	AGG	Arg	AAG	Lys	ACG	Thr	AUG	Met	G
	AGA	Arg	AAA	Lys	ACA	Thr	AUA	Iso	A
	AGC	Ser	AAC	Asn	ACC	Thr	AUC	Iso	C
	AGU	Ser	AAU	Asn	ACU	Thr	AUU	Iso	U
C	CGG	Arg	CAG	Gln	CCG	Pro	CUG	Leu	G
	CGA	Arg	CAA	Gln	CCA	Pro	CUA	Leu	A
	CGC	Arg	CAC	Hist	CCC	Pro	CUC	Leu	C
	CGU	Arg	CAU	Hist	CCU	Pro	CUU	Leu	U
U	UGG	Trp	UAG	stop	UCG	Ser	UUG	Leu	G
	UGA	stop	UAA	stop	UCA	Ser	UUA	Leu	A
	UGC	Cyst	UAC	Tyr	UCC	Ser	UUC	Phe	C
	UGU	Cyst	UAU	Tyr	UCU	Ser	UUU	Phe	U

Suggest **one** advantage of showing the genetic code as base sequences on mRNA, rather than triplets on DNA.

(1)

(c) What name is given to a group of three bases on mRNA that codes for an amino acid?

(1)

(d) Use information from **Table 1** to explain why the genetic code is described as degenerate.

(2)

(e) Suggest the role of the mRNA base triplets UGA, UAG and UAA.

(2)

(f) **Table 2** shows the sequence of mRNA bases forming part of a single gene.

Table 2

Base on DNA template									
Base on mRNA	G	U	G	U	A	C	U	G	G
Encoded amino acid									

Complete **Table 2** to show the base sequence of the DNA template from which this mRNA was transcribed and the encoded amino acid sequence.

(2)

(Total 10 marks)

Q3. (a) Describe the cohesion-tension theory of water transport in the xylem.

(b) Identify the molecule labelled **Q**.

(1)

(c) In the diagram above, the first codon is AUG. Give the base sequence of:

the complementary DNA base sequence _____

the missing anticodon _____

(2)

The table below shows the base triplets that code for two amino acids.

Amino acid	Encoding base triplet
Aspartic acid	GAC, GAU
Proline	CCA, CCG, CCC, CCU

(d) Aspartic acid and proline are both amino acids. Describe how two amino acids differ from one another. You may use a diagram to help your description.

(1)

(e) Deletion of the sixth base (G) in the sequence shown in the diagram above would change the nature of the protein produced but substitution of the same base would not. Use the information in the table and your own knowledge to explain why.

(Extra space) _____

(3)

(Total 8 marks)

Q5. Haemoglobin transports oxygen around the body of many animals.

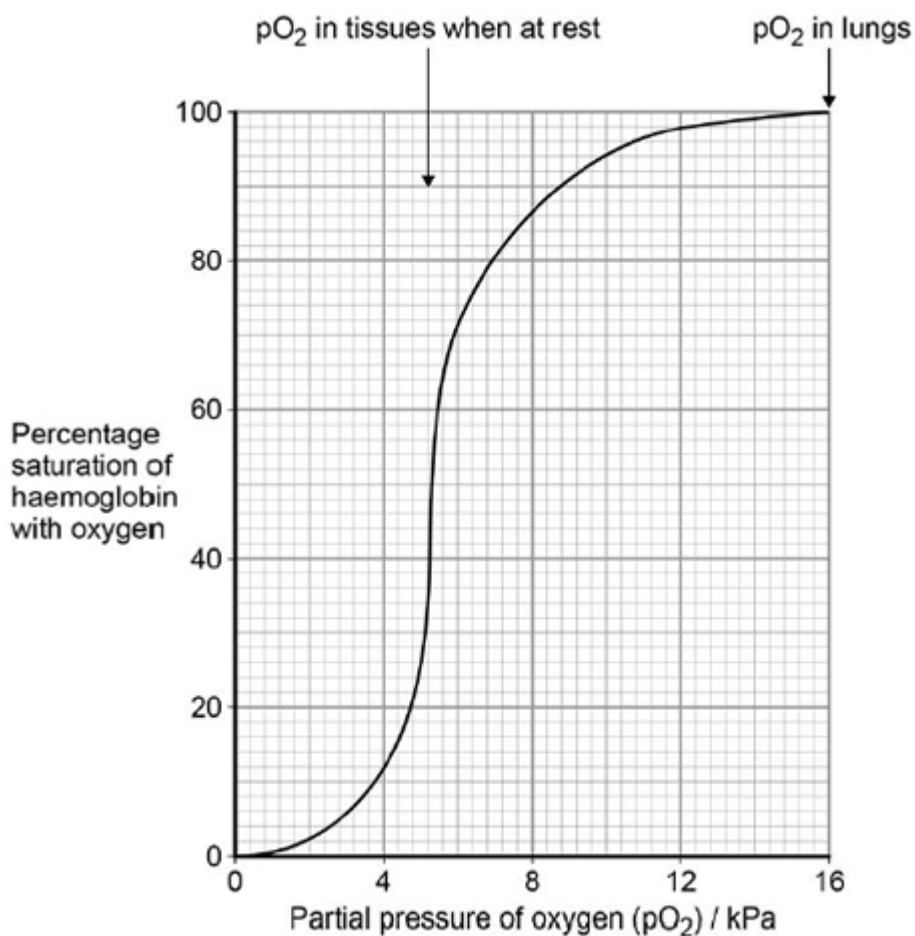
(a) Haemoglobin is a protein with a quaternary structure.

Explain the meaning of **quaternary structure**.

(1)

(b) When fully saturated, each molecule of haemoglobin is bound to four molecules of oxygen.

The graph shows the percentage saturation of haemoglobin with oxygen at different partial pressures.



Give the formula for calculating the percentage saturation of haemoglobin with oxygen.

Percentage
saturation of
haemoglobin with
oxygen =

(1)

- (c) The first molecule of oxygen to bind causes a change in the shape of the haemoglobin molecule.

This change of shape makes it easier for other oxygen molecules to bind to the haemoglobin molecule.

Explain how the graph provides evidence for this.

(2)

- (d) Suggest **one** advantage of this change in the affinity of haemoglobin for oxygen.

(1)

- (e) Tests on the man whose blood was used to construct the graph gave the following data.

- Concentration of haemoglobin in blood = 150 g dm^{-3} .
- Volume of oxygen carried by fully saturated haemoglobin = $1.35 \text{ cm}^3 \text{ g}^{-1}$.
- Resting heart rate = $60 \text{ beats minute}^{-1}$.
- Volume of blood pumped out of left ventricle each beat = 60 cm^3 .

Use these data and information from the graph to calculate the volume of oxygen released to the man's tissues per minute whilst he was at rest.

Show your working.

Answer = _____ $\text{cm}^3 \text{ minute}^{-1}$

(3)

(Total 8 marks)

Q6. In a eukaryotic cell, transcription results in a molecule of pre-mRNA that is modified to produce mRNA. In a prokaryotic cell transcription produces mRNA directly.

(a) Explain this difference.

(2)

(b) Give **two** differences between the structure of mRNA and the structure of tRNA.

1. _____

2. _____

(2)

(Total 4 marks)

Q7. (a) Give the **two** types of molecule from which a ribosome is made.

(1)

(b) Describe the role of a ribosome in the production of a polypeptide. Do **not** include transcription in your answer.

(3)

(c) The table below shows the base sequence of part of a pre-mRNA molecule from a eukaryotic cell.

Complete the table with the base sequence of the DNA strand from which this pre-mRNA

was transcribed.

									DNA
A	C	G	C	A	U	U	A	U	pre-mRNA

(1)

- (d) In a eukaryotic cell, the base sequence of the mRNA might be different from the sequence of the pre-mRNA.

Explain why.

(2)

(Total 7 marks)

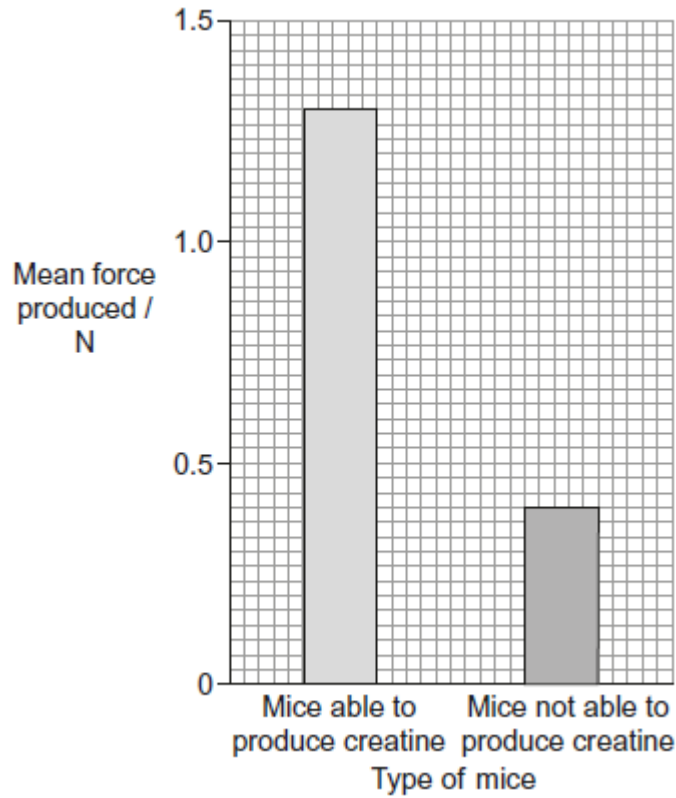
- Q8.** (a) What is the role of ATP in myofibril contraction?

(2)

- (b) Scientists investigated the effect of not being able to produce creatine on the force produced by muscle. They used mice with a mutation that made them not able to produce creatine.

The force produced when these mice gripped with their paws was compared with the force produced by normal mice that were able to produce creatine.

The graph shows the scientists' results.



- (i) What was the percentage fall in the mean force produced by mice not able to produce creatine, compared with the normal mice? Show your working.

Answer _____ % (2)

- (ii) Suggest an explanation for these results.

(2)

- (c) The mice that were not able to produce creatine were homozygous for a recessive allele of a gene. Mice that are heterozygous for this allele are able to produce forces similar to those of normal mice that are homozygous for the dominant allele of the same gene.

Explain why the heterozygous mice can produce forces similar to those of normal mice.

(2)

(Total 8 marks)

Q9. Haemoglobin is a protein. It is made of two alpha polypeptides and two beta polypeptides. Each alpha polypeptide has 141 amino acids and each beta polypeptide has 146 amino acids.

- (a) What term is used to describe the structure of a protein made of two or more polypeptides?

(1)

- (b) Calculate the minimum number of DNA bases needed to code for the number of amino acids in one alpha polypeptide.

Answer = _____

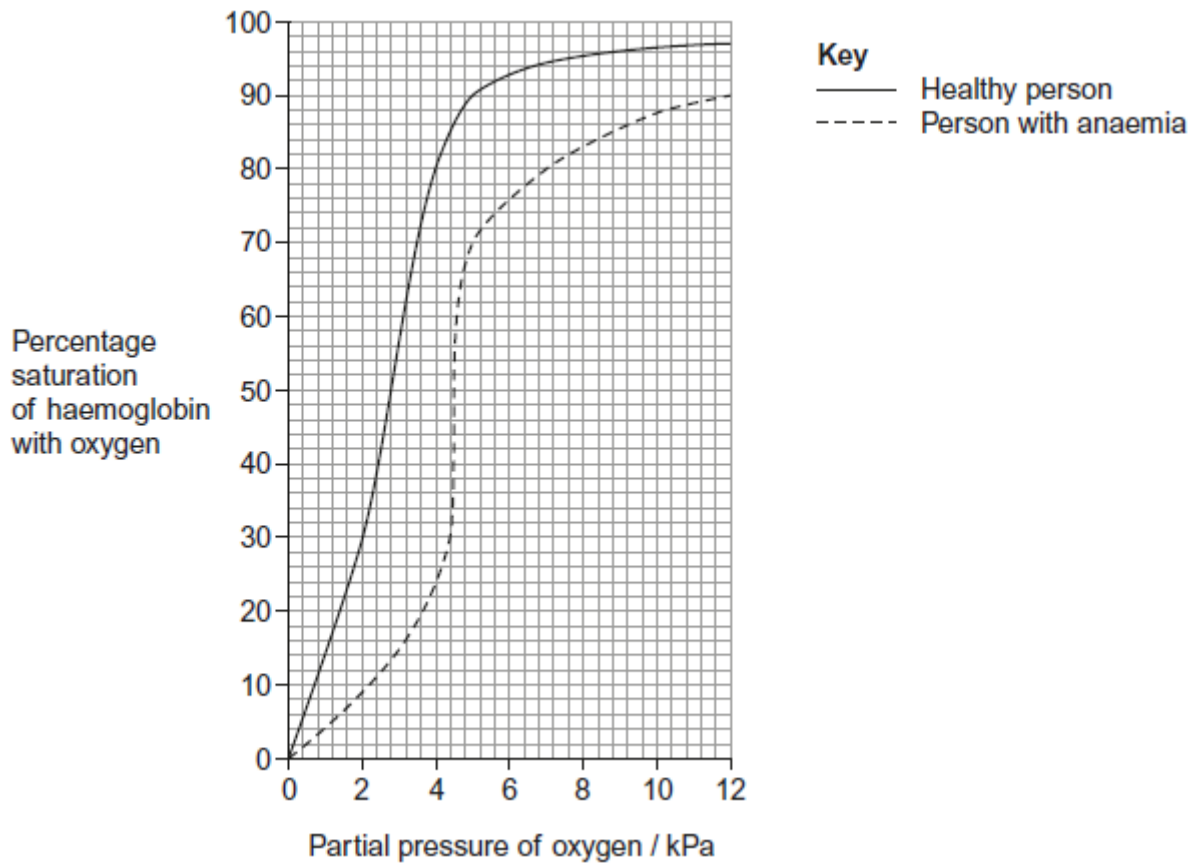
(1)

- (c) Describe the role of haemoglobin in supplying oxygen to the tissues of the body.

(2)

Anaemia is a condition in which there is a decrease in the concentration of haemoglobin in blood. In some people with anaemia, substances are produced which change the oxygen dissociation curve of haemoglobin.

The graph shows the effect of these substances on the oxygen dissociation curve of haemoglobin.



- (d) (i) Use information in the graph to find the difference in the percentage saturation of haemoglobin with oxygen between a healthy person and a person with anaemia at a partial pressure of oxygen of 4 kPa.

Answer = _____ (1)

- (ii) Explain the advantage to a person with anaemia of the change shown in the oxygen dissociation curve.

(3)

Q10. Read the following passage carefully.

A large and growing number of disorders are now known to be due to types of mitochondrial disease (MD). MD often affects skeletal muscles, causing muscle weakness.

We get our mitochondria from our mothers, via the fertilised egg cell. Fathers do not pass on mitochondria via their sperm. Some mitochondrial diseases are caused by mutations of mitochondrial genes inside the mitochondria. Most mitochondrial diseases are caused by mutations of genes in the cell nucleus that are involved in the functioning of mitochondria. These mutations of nuclear DNA produce recessive alleles. 5

One form of mitochondrial disease is caused by a mutation of a mitochondrial gene that codes for a tRNA. The mutation involves substitution of guanine for adenine in the DNA base sequence. This changes the anticodon on the tRNA. This results in the formation of a non-functional protein in the mitochondrion. 10

There are a number of ways to try to diagnose whether someone has a mitochondrial disease. One test involves measuring the concentration of lactate in a person's blood after exercise. In someone with MD, the concentration is usually much higher than normal. If the lactate test suggests MD, a small amount of DNA can be extracted from mitochondria and DNA sequencing used to try to find a mutation. 15

20

Use information in the passage and your own knowledge to answer the following questions.

- (a) Mitochondrial disease (MD) often causes muscle weakness (lines 1–3). Use your knowledge of respiration and muscle contraction to suggest explanations for this effect of MD.

(Extra space) _____

(3)

Two couples, couple **A** and couple **B**, had one or more children affected by a mitochondrial disease. The type of mitochondrial disease was different for each couple.

None of the parents showed signs or symptoms of MD.

- Couple **A** had four children who were all affected by an MD.
- Couple **B** had four children and only one was affected by an MD.

(b) Use the information in lines 5–9 and your knowledge of inheritance to suggest why:

- all of couple **A**'s children had an MD
- only one of couple **B**'s children had an MD.

Couple **A** _____

Couple **B** _____

(Extra space) _____

(4)

(c) Suggest how the change in the anticodon of a tRNA leads to MD (lines 10–13).

(Extra space) _____

(3)

(d) If someone has MD, the concentration of lactate in their blood after exercise is usually much higher than normal (lines 15–17). Suggest why.

(Extra space) _____

(3)

(e) A small amount of DNA can be extracted from mitochondria and DNA sequencing used to try to find a mutation (lines 18–19).

From this sample:

- how would enough DNA be obtained for sequencing?
- how would sequencing allow the identification of a mutation?

(2)

(Total 15 marks)

Q11. (a) Explain how the structure of DNA is related to its functions.

(Extra space)

(6)

Scientists investigated three genes, **C**, **D** and **E**, involved in controlling cell division. They studied the effect of mutations in these genes on the risk of developing lung cancer.

The scientists analysed genes **C**, **D** and **E** from healthy people and people with lung cancer.

- If a person had a normal allele for a gene, they used the symbol N.
- If a person had two mutant alleles for a gene, they used the symbol M.

They used their data to calculate the risk of developing lung cancer for people with different combinations of N and M alleles of the genes. A risk value of 1.00 indicates no increased risk. The following table shows the scientists' results.

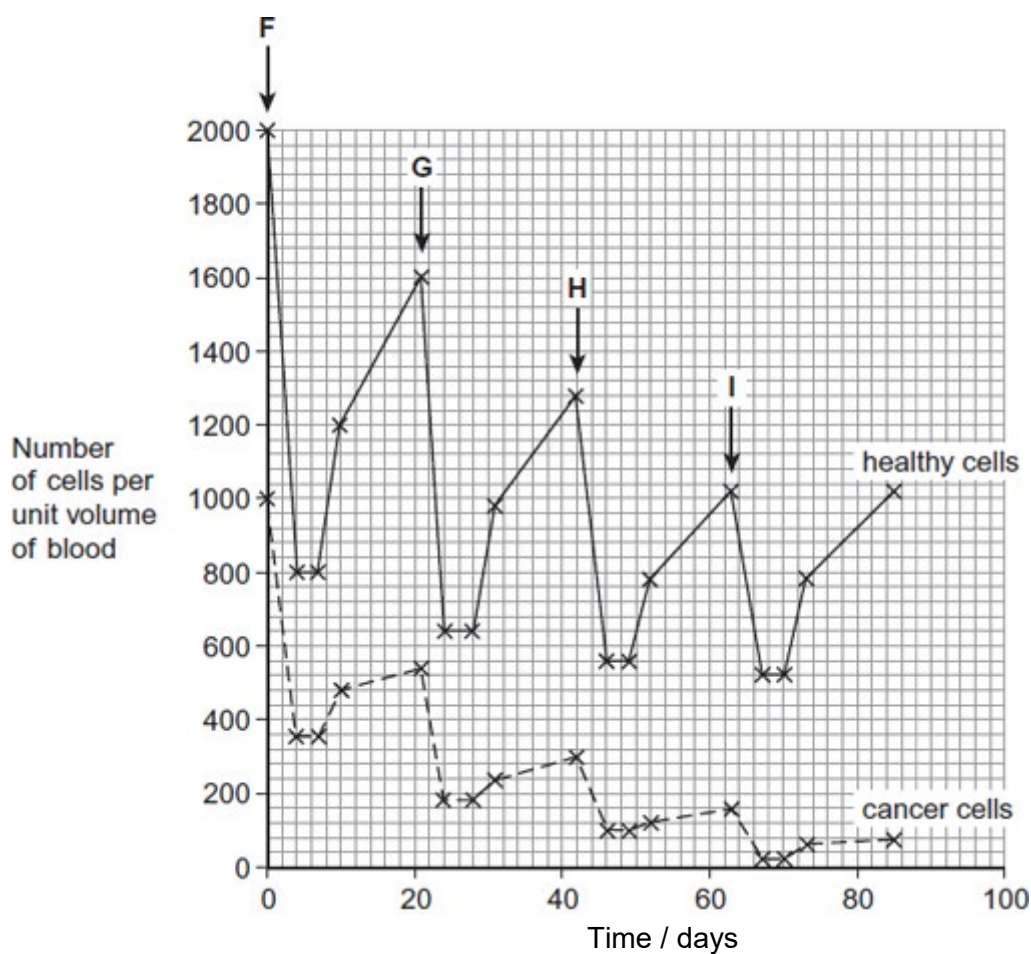
Gene C	Gene D	Gene E	Risk of developing lung cancer
N	N	N	1.00
M	N	N	1.30
N	N	M	1.78
N	M	N	1.45

N = at least one copy of the normal allele is present
M = two copies of the mutant allele are present

- (b) What do these data suggest about the relative importance of the mutant alleles of genes **C**, **D** and **E** on **increasing** the risk of developing lung cancer? Explain your answer.

(3)

Chemotherapy is the use of a drug to treat cancer. The drug kills dividing cells. The figure below shows the number of healthy cells and cancer cells in the blood of a patient receiving chemotherapy. The arrows labelled F to I show when the drug was given to the patient.



(c) Calculate the rate at which healthy cells were killed between days 42 and 46.

_____ cells killed per unit volume of blood per day (1)

- (d) Describe similarities and differences in the response of healthy cells and cancer cells to the drug between times **F** and **G**.

(Extra space) _____

(3)

- (e) More cancer cells could be destroyed if the drug was given more frequently.
Suggest why the drug was **not** given more frequently.

(2)

(Total 15 marks)

- Q12.** (a) (i) Why is the genetic code described as being universal?

(1)

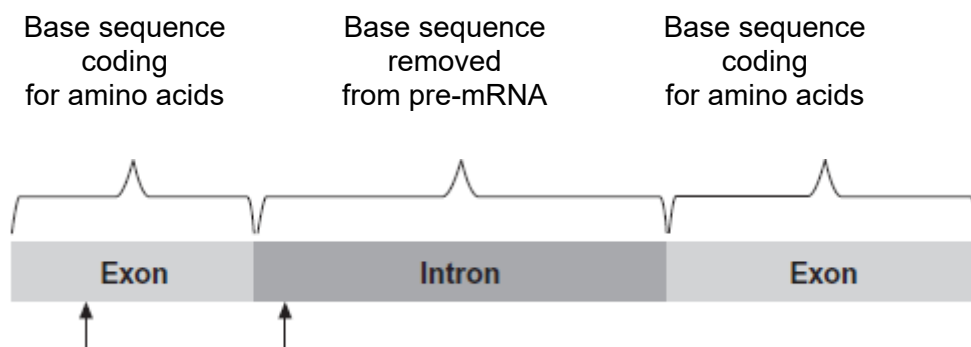
- (ii) The genetic code uses four different DNA bases. What is the maximum number of different DNA triplets that can be made using these four bases?

(1)

Transcription of a gene produces pre-mRNA.

- (b) Name the process that removes base sequences from pre-mRNA to form mRNA.

- (c) The figure below shows part of a pre-mRNA molecule. Geneticists identified two mutations that can affect this pre-mRNA, as shown in the figure.



Mutation 1, single base deletion **Mutation 2, single base substitution**

- (i) **Mutation 1** leads to the production of a non-functional protein.

Explain why.

(Extra space) _____

(3)

- (ii) What effect might **mutation 2** have on the protein produced?

Explain your answer.

(2)

(Total 8 marks)

acid missing.

(2)

- (ii) Suggest how the production of a protein with one amino acid missing may lead to a genetic disorder such as Ellis-van Creveld syndrome.

(2)

(Total 5 marks)

- Q15.** (a) The genetic code is described as being degenerate. What does this mean?

(1)

- (b) What is a codon?

(2)

- (c) (i) What is the role of RNA polymerase during transcription?

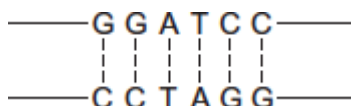
(1)

(ii) mRNA can be converted to cDNA.

Name the enzyme used in this process.

(1)

(d) The diagram shows the base sequence on DNA where a restriction endonuclease cuts DNA.



Use evidence from the diagram to explain what is meant by a palindromic recognition sequence on DNA.

(1)

(Total 6 marks)

Q16. (a) (i) A mutation of a tumour suppressor gene can result in the formation of a tumour.

Explain how.

(2)

(ii) Not all mutations result in a change to the amino acid sequence of the encoded polypeptide.

Explain why.

(1)

(b) Some cancer cells have a receptor protein in their cell-surface membrane that binds to a

hormone called **growth factor**. This stimulates the cancer cells to divide.

Scientists have produced a monoclonal antibody that stops this stimulation.

Use your knowledge of monoclonal antibodies to suggest how this antibody stops the growth of a tumour.

[Extra space] _____

(3)

(Total 6 marks)

Q17. The black mamba is a poisonous snake. Its poison contains a toxin.

The table shows the base sequence of mRNA that codes for the first two amino acids of this toxin.

Base sequence of anticodon on tRNA						
Base sequence of mRNA	A	C	G	A	U	G
Base sequence of DNA						

Complete the table to show

(a) (i) the base sequence of the anticodon on the first tRNA molecule that would bind to this mRNA sequence

(1)

(ii) the base sequence of the DNA from which this mRNA was transcribed.

(1)

(b) The length of the section of DNA that codes for the complete toxin is longer than the mRNA used for translation. Explain why.

(1)

- (c) A mutation in the base sequence of the DNA that codes for the toxin would change the base sequence of the mRNA.

Explain how a change in the base sequence of the mRNA could lead to a change in the tertiary structure of the toxin.

(1)

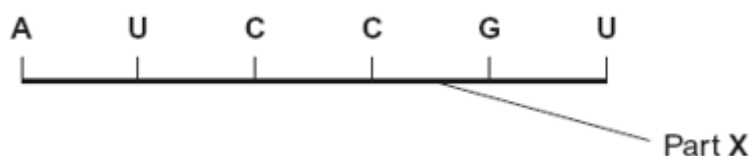
- (d) The black mamba's toxin kills prey by preventing their breathing. It does this by inhibiting the enzyme acetylcholinesterase at neuromuscular junctions. Explain how this prevents breathing.

(Extra space) _____

(3)

(Total 7 marks)

Q18. The diagram shows part of a pre-mRNA molecule.



- (a) (i) Name the **two** substances that make up part X.

_____ and _____

(1)

- (ii) Give the sequence of bases on the DNA strand from which this pre-mRNA has been transcribed.

(1)

- (b) (i) Give one way in which the structure of an mRNA molecule is different from the structure of a tRNA molecule.

(1)

- (ii) Explain the difference between pre-mRNA and mRNA.

(1)

- (c) The table shows the percentage of different bases in two pre-mRNA molecules. The molecules were transcribed from the DNA in different parts of a chromosome.

Part of chromosome	Percentage of base			
	A	G	C	U
Middle	38	20	24	
End	31	22	26	

- (i) Complete the table by writing the percentage of uracil (U) in the appropriate boxes.

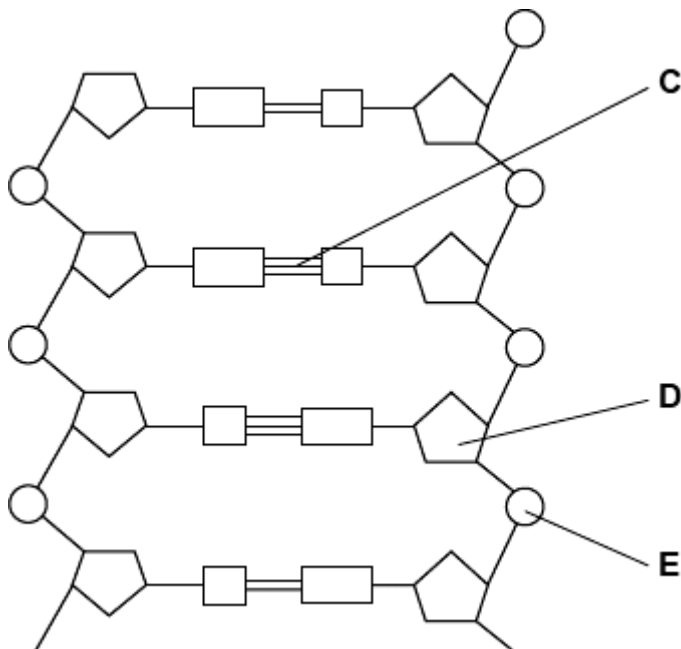
(1)

- (ii) Explain why the percentages of bases from the middle part of the chromosome and the end part are different.

(2)

(Total 7 marks)

Q19. The diagram shows part of a DNA molecule.



(a) (i) DNA is a polymer. What is the evidence from the diagram that DNA is a polymer?

(1)

(ii) Name the parts of the diagram labelled **C**, **D** and **E**.

Part **C** _____

Part **D** _____

Part **E** _____

(3)

(iii) In a piece of DNA, 34% of the bases were thymine.

Complete the table to show the names and percentages of the other bases.

Name of base	Percentage
Thymine	34
	34

(2)

(b) A polypeptide has 51 amino acids in its primary structure.

(i) What is the minimum number of DNA bases required to code for the amino acids in

this polypeptide?

(1)

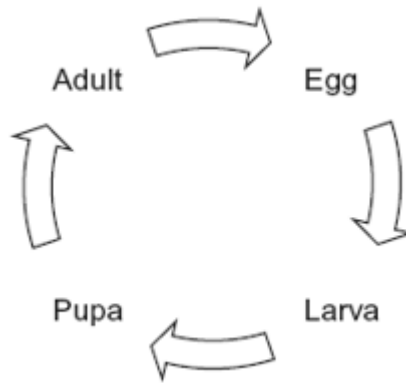
(ii) The gene for this polypeptide contains more than this number of bases.

Explain why

(1)

(Total 8 marks)

Q20. The diagram shows the life cycle of a fly.



When the larva is fully grown, it changes into a pupa. The pupa does not feed. In the pupa, the tissues that made up the body of the larva are broken down. New adult tissues are formed from substances obtained from these broken-down tissues and from substances that were stored in the body of the larva.

(a) Hydrolysis and condensation are important in the formation of new adult proteins. Explain how.

(2)

(b) Most of the protein stored in the body of a fly larva is a protein called calliphorin. Explain why different adult proteins can be made using calliphorin.

(1)

The table shows the mean concentration of RNA in fly pupae at different ages.

Age of pupa as percentage of total time spent as a pupa	Mean concentration of RNA / μg per pupa
0	20
20	15
40	12
60	17
80	33
100	20

(c) Describe how the concentration of RNA changes during the time spent as a pupa.

(2)

(d) (i) Describe how you would expect the number of lysosomes in a pupa to change with the age of the pupa. Give a reason for your answer.

(2)

(ii) Suggest an explanation for the change in RNA concentration in the first 40% of the time spent as a pupa.

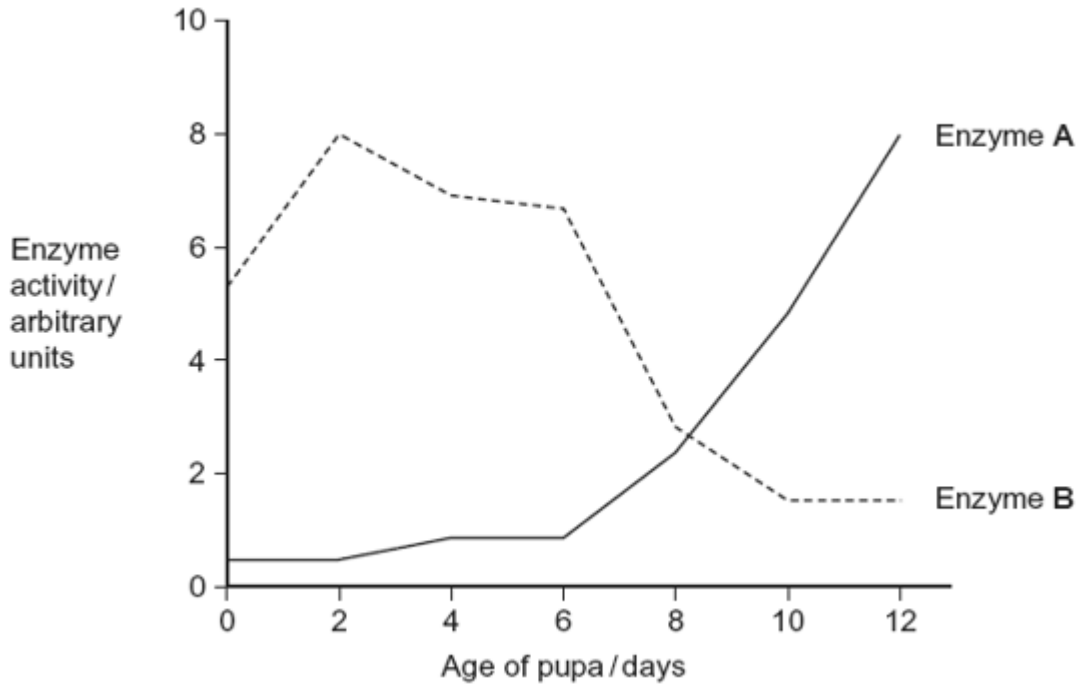
(2)

(e) Suggest an explanation for the change in RNA concentration between 60 and 80% of the time spent as a pupa.

(2)

(f) The graph shows changes in the activity of two respiratory enzymes in a fly pupa.

- Enzyme **A** catalyses a reaction in the Krebs cycle
- Enzyme **B** catalyses the formation of lactate from pyruvate



During the first 6 days as a pupa, the tracheae break down. New tracheae are formed after 6 days. Use this information to explain the change in activity of the two enzymes.

(Extra space) _____

(4)

(Total 15 marks)

Q21. CREB is a transcription factor in the mitochondria of neurones.

(a) What is a **transcription factor**?

(2)

(b) CREB leads to the formation of a protein that removes electrons and protons from reduced NAD in the mitochondrion.

Huntington's disease (HD) causes the death of neurones. People with HD produce a substance called huntingtin. Some scientists have suggested that binding of huntingtin to CREB may lead to the death of neurones.

Suggest how binding of huntingtin to CREB may lead to the death of neurones.

(Extra space) _____

(3)

(c) CREB is a protein synthesised in the cytoplasm of neurones. Transport of CREB from the cytoplasm into the matrix of a mitochondrion requires two carrier proteins.

Use your knowledge of the structure of a mitochondrion to explain why transport of CREB requires **two** carrier proteins.

(2)

(Total 7 marks)

Q22. The diagram shows a short sequence of DNA bases.

TTTGTATACTAGTCTACTTCGTTAATA

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

(1)

- (ii) The number of amino acids coded for could be fewer than your answer to part (a)(i).

Give **one** reason why.

(1)

- (b) Explain how a change in the DNA base sequence for a protein may result in a change in the structure of the protein.

(Extra space) _____

(3)

- (c) A piece of DNA consisted of 74 base pairs. The two strands of the DNA, strands **A** and **B**, were analysed to find the **number** of bases of each type that were present. Some of the results are shown in the table.

	Number of bases			
	C	G	A	T
Strand A	26			
Strand B	19		9	

Complete the table by writing in the missing values.

(2)
(Total 7 marks)

Q23. (a) What name is used for the non-coding sections of a gene?

(1)

Figure 1 shows a DNA base sequence. It also shows the effect of two mutations on this base sequence. **Figure 2** shows DNA triplets that code for different amino acids.

Figure 1

Original DNA base sequence	A	T	T	G	G	C	G	T	G	T	C	T
Amino acid sequence												
Mutation 1 DNA base sequence	A	T	T	G	G	A	G	T	G	T	C	T
Mutation 2 DNA base sequence	A	T	T	G	G	C	C	T	G	T	C	T

Figure 2

DNA triplets	Amino acid
GGT, GGC, GGA, GGG	Gly
GTT, GTA, GTG, GTC	Val
ATC, ATT, ATA	Ile
TCC, TCT, TCA, TCG	Ser
CTC, CTT, CTA, CTG	Leu

(b) Complete **Figure 1** to show the sequence of amino acids coded for by the original DNA base sequence.

(1)

(c) Some gene mutations affect the amino acid sequence. Some mutations do not. Use the information from **Figure 1** and **Figure 2** to explain

(i) whether mutation 1 affects the amino acid sequence

(2)

(ii) how mutation 2 could lead to the formation of a non-functional enzyme.

(3)

(d) Gene mutations occur spontaneously.

(i) During which part of the cell cycle are gene mutations most likely to occur?

(1)

(ii) Suggest an explanation for your answer.

(1)

(Total 9 marks)

Q24. (a) The table below shows features of a bacterium and the human immunodeficiency virus (HIV) particle.

Complete the table by putting a tick (✓) where a feature is present.

Feature	Bacterium	Human immunodeficiency virus (HIV) particle
RNA		
Cell wall		
Enzyme molecules		
Capsid		

(2)

(b) When HIV infects a human cell, the following events occur.

- A single-stranded length of HIV DNA is made.
- The human cell then makes a complementary strand to the HIV DNA.

The complementary strand is made in the same way as a new complementary strand is made during semi-conservative replication of human DNA.

Describe how the complementary strand of HIV DNA is made.

The scientists collected data from each cheetah on four separate occasions. **Figure 2** shows the data for one of the cheetahs.

Figure 2

Side of tail	Mean band width score (\pm standard deviation)						
	Band 1	Band 2	Band 3	Band 4	Band 5	Band 6	Band 7
Right	3.00 (\pm 0.82)	1.00 (\pm 0.00)	1.00 (\pm 0.00)	3.75 (\pm 0.50)	2.75 (\pm 0.50)	3.00 (\pm 0.00)	3.00 (\pm 0.00)
Left	3.75 (\pm 0.50)	3.25 (\pm 0.50)	2.00 (\pm 0.50)	3.00 (\pm 0.00)	2.00 (\pm 0.00)	2.50 (\pm 0.50)	3.00 (\pm 0.50)

(a) The scientists only used data from cheetahs which were fully grown. Suggest why.

(1)

(b) The scientists estimated the width of the bands on the same cheetah on four separate occasions. They did not always get the same score.

(i) Give **two** pieces of evidence from **Figure 2** which show that the scientists sometimes obtained different scores for the same band.

1. _____

2. _____

(2)

(ii) The method the scientists used resulted in them getting different scores for the same band. Suggest why.

(1)

(c) What is the evidence from **Figure 2** that the dark and light bands do **not** form rings of equal width around the tail?

(1)

(d) The scientists found the difference in banding pattern between

- offspring in the same family
- cheetahs chosen randomly.

Explain how scientists could use this information to show that some variation in tail banding was genetic.

(Extra space) _____

(3)

(Total 8 marks)

Q26. (a) The genetic code is **degenerate** and **non-overlapping**.

Explain the meaning of:

Degenerate _____

Non-overlapping _____

(2)

The table shows a short section of a messenger RNA (mRNA) molecule and the section of a polypeptide for which it codes.

mRNA	G G G	G C U	U C A	C C G	G C A	A C G
Polypeptide	glycine	alanine	serine	proline	alanine	threonine

(b) Name the bases represented in the table by:

A _____

C _____

G _____

U _____

(2)

(c) Use information in the table to give the sequence of bases in **DNA** that codes for serine.

(1)

(Total 5 marks)

