

DNA and Chromosomes

These practice questions can be used by students and teachers and is

Suitable for AQA A Level 7402 Biology Topic Question

Level: AQA A LEVEL 7402

Subject: Biology

Exam Board: AQA A Level 7402

Topic: DNA and Chromosomes

1

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)

2

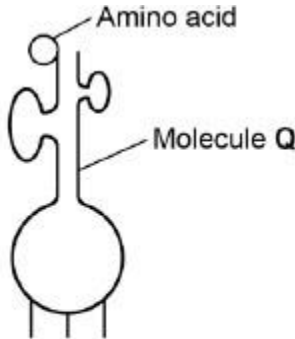
The diagram below represents one process that occurs during protein synthesis.

(a) Name the process shown.

(1)

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

(1)



For more hel

A U G C C G U A C C G A C U

- (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)



(c) Describe how proteins are digested in the human gut.

(4)

(Total 15 marks)

4

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) _____

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)

5

- (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?

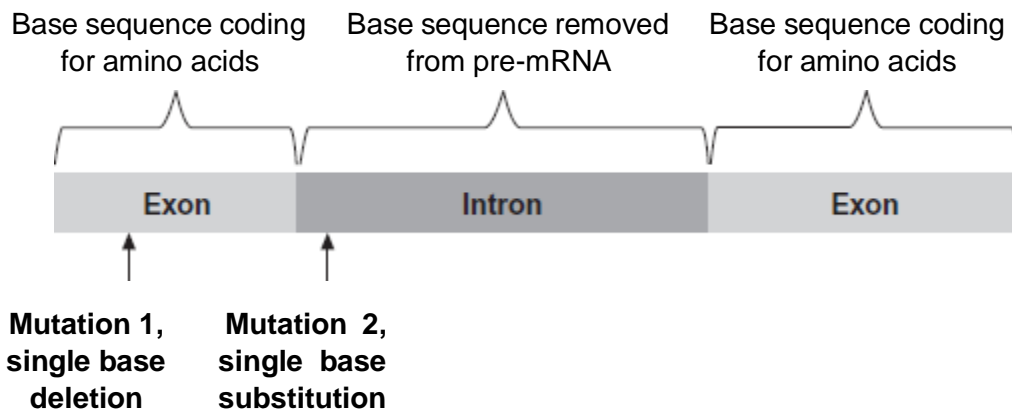
(1)

Transcription of a gene produces pre-mRNA.

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

(1)

- (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.



- (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)

6

(a) What is the name of a _____ position of a gene on a chromosome?

(1)

(b) What is meant by genetic diversity?

(1)

A geneticist investigated genetic diversity in four different breeds of dog. She compared DNA base sequences of the same genes from a large number of dogs from each breed.

The geneticist calculated the mean genetic diversity for each breed of dog. The value of this mean was between 0 and 1.

- A mean value of 1 shows maximum genetic diversity.
- A mean value of 0 shows no genetic diversity.

Her results are shown in the table

| Breed of dog | Mean genetic diversity | Standard deviation |
|----------------------|------------------------|--------------------|
| Airedale terrier | 0.51 | ± 0.03 |
| Bull terrier | 0.38 | ± 0.02 |
| Jack Russell terrier | 0.76 | ± 0.01 |
| Miniature terrier | 0.47 | ± 0.02 |

(c) What do these data show about the differences in genetic diversity between these breeds of dog?

(3)

(d) Miniature terriers were _____ first bred from bull terriers in the 19th century.

Suggest **one** explanation for the observed difference in genetic diversity between miniature terriers and bull terriers.

(2)

(Total 7 marks)

7

The Amish are a group of people who live in America. This group was founded by 30 Swiss people, who moved to America many years ago. The Amish do not usually marry people from outside their own group.

One of the 30 Swiss founders had a genetic disorder called Ellis-van Creveld syndrome. People with this disorder have heart defects, are short and have extra fingers and toes. Ellis-van Creveld syndrome is caused by a faulty allele.

In America today, about 1 in 200 Amish people are born with Ellis-van Creveld syndrome. This disorder is very rare in people in America who are not Amish.

(a) In America today, there are approximately 1250 Amish people who have Ellis-van Creveld syndrome. Use the information provided to calculate the current Amish population of America.

Amish population _____

(1)



(b) The faulty allele that _____ causes Ellis-van Creveld syndrome is the result of a mutation of a gene called *EVC*. This mutation leads to the production of a protein that has one aminoacid missing.

(i) Suggest how a mutation can lead to the production of a protein that has one amino 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)

8

(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?

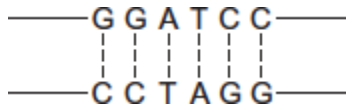
(1)

(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)

9

Phenylketonuria is a disease caused by mutations of the gene coding for the enzyme PAH. The table shows part of the DNA base sequence coding for PAH. It also shows a mutation of this sequence which leads to the production of non-functioning PAH.

| | | | | | | | | | | | | |
|--|---|---|---|---|---|---|---|---|---|---|---|---|
| DNA base sequence coding for PAH | C | A | G | T | T | C | G | C | T | A | C | G |
| DNA base sequence coding for non-functioning PAH | C | A | G | T | T | C | C | C | T | A | C | G |

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

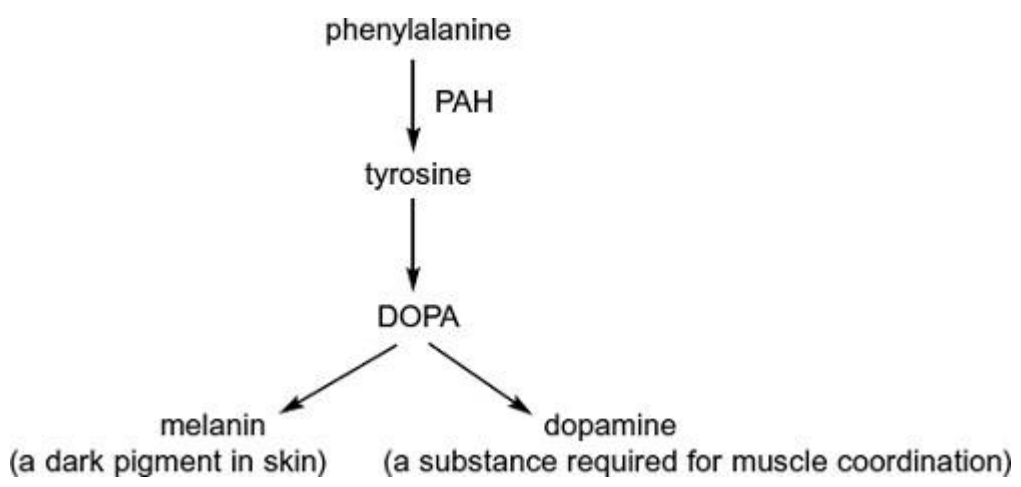
(1)

(ii) Explain how this mutation leads to the formation of non-functioning PAH.

(Extra space)

(3)

PAH catalyses a reaction at the start of two enzyme-controlled pathways. The diagram shows these pathways.



(b) Use the information in the diagram to give **two** symptoms you might expect to be visible in a person who produces non-functioning PAH.

1. _____

2. _____

(2)

- (c) One mutation causing _____ phenylketonuria was originally only found in one population in central Asia. It is now found in many different populations across Asia. Suggest how the spread of this mutation may have occurred.

(1)

10

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.

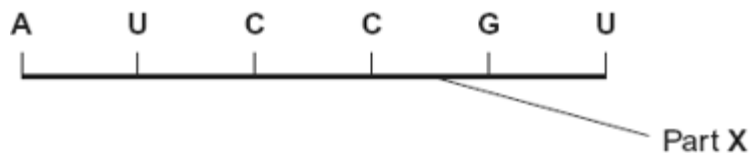
| | Number of bases | | | |
|-----------------|-----------------|---|---|---|
| | C | G | A | T |
| Strand A | 26 | | | |
| Strand B | 19 | | 9 | |

Complete the table by writing in the missing values.

(2)
(Total 7 marks)

11

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.
- _____
- _____
- (3)

- (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)

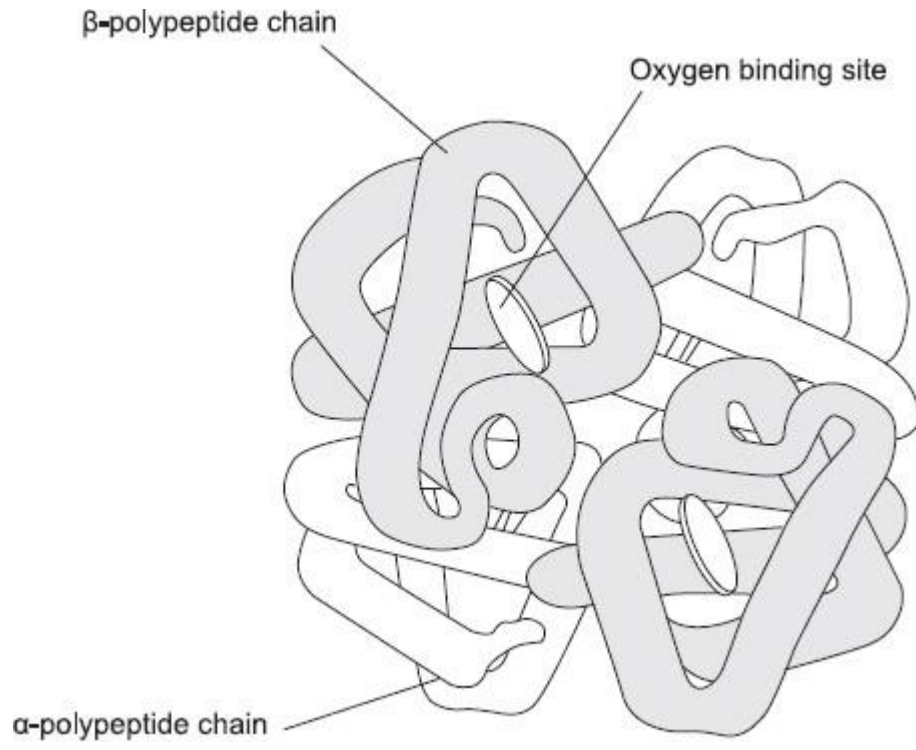
- (ii) Explain why the percentages of bases from the middle part of the chromosome and the end part are different.

(2)

(Total 7 marks)

12

The diagram shows a molecule _____ of haemoglobin.



(a) What is the evidence from the diagram that haemoglobin has a quaternary structure?

(1)

(b) (i) A gene codes for the α -polypeptide chain. There are 423 bases in this gene that code for amino acids. How many amino acids are there in the α -polypeptide chain?

(1)

(ii) The total number of bases in the DNA of the α -polypeptide gene is more than 423.

Give **two** reasons why there are more than 423 bases.

1. _____

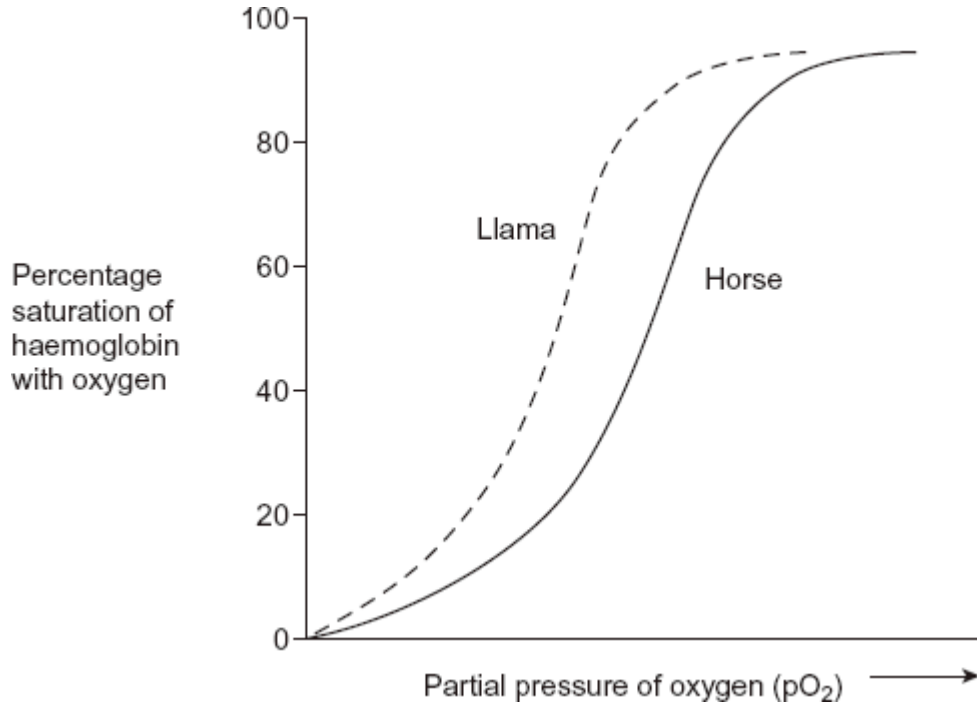
2. _____

(2)

- (c) The haemoglobin in one organism may have a different chemical structure from the haemoglobin in another organism. Describe how.

(1)

- (d) The graph shows oxygen dissociation curves for horse haemoglobin and for llama haemoglobin. Horses are adapted to live at sea level and llamas are adapted to live in high mountains.



Use the graph to explain why llamas are better adapted to live in high mountains than horses.

(3)
(Total 8 marks)

13

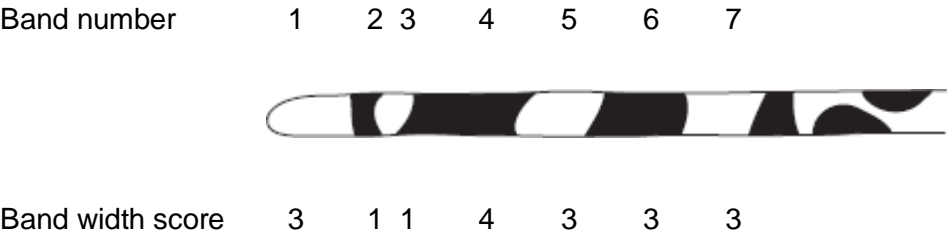
The body markings of cheetahs vary, in particular the pattern of bands on their tails. Cheetahs are solitary animals but the young stay with their mother until they are between 14 and 18 months old.

Scientists investigated the banding pattern on the tails of cheetahs living in the wild.

- They drove a car alongside a walking cheetah and used binoculars to study the tail pattern.
- They gave each cheetah a banding pattern score based on the width of the dark and light bands on the end of the tail.
- They scored the width of the bands on the right and left side of the tail using a 5 point scale of width.

A typical pattern on the right side of one cheetah’s tail is shown in **Figure 1**.

Figure 1



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)

14

- (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)

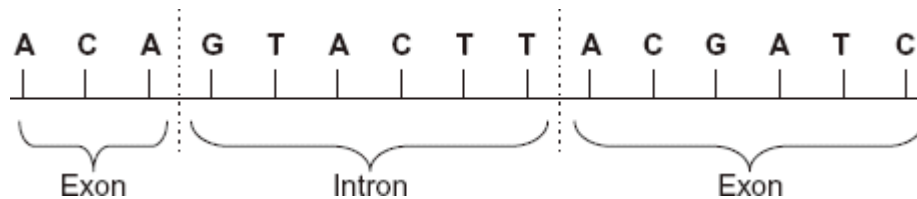
15

(a) Complete the table to show the differences between DNA, mRNA and tRNA.

| Type of nucleic acid | Hydrogen bonds present (✓) or not present (✗) | Number of polynucleotide strands in molecule |
|----------------------|---|--|
| DNA | | |
| mRNA | | |
| tRNA | | |

(2)

(b) The diagram shows the bases on one strand of a piece of DNA.



(i) In the space below, give the sequence of bases on the pre-mRNA transcribed from this strand.

(2)

(ii) In the space below, give the sequence of bases on the mRNA produced by splicing this piece of pre-mRNA.

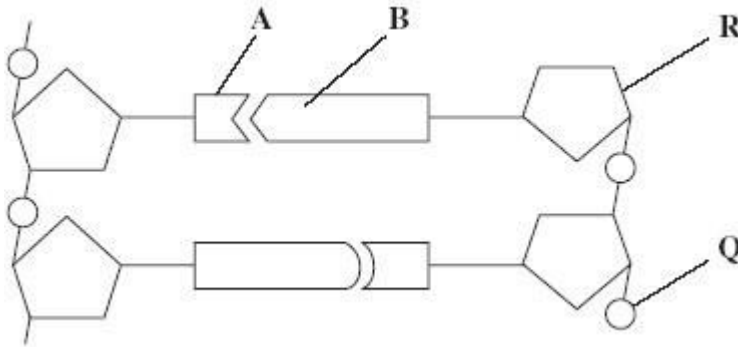
(1)

(Total 5 marks)

16

Figure 1 shows a short section of a DNA molecule.

Figure 1



(a) Name parts **R** and **Q**.

(i) **R** _____

(ii) **Q** _____

(2)

(b) Name the bonds that join **A** and **B**.

(1)

(c) Ribonuclease is an enzyme. It is 127 amino acids long.

What is the minimum number of DNA bases needed to code for ribonuclease?

(1)

- (d) **Figure 2** shows the sequence of DNA bases coding for seven amino acids in the enzyme ribonuclease.

Figure 2

G T T T A C T A C T C T T C T T C T T T A

The number of each type of amino acid coded for by this sequence of DNA bases is shown in the table.

| Amino acid | Number present |
|------------|----------------|
| Arg | 3 |
| Met | 2 |
| Gln | 1 |
| Asn | 1 |

Use the table and **Figure 2** to work out the sequence of amino acids in this part of the enzyme. Write your answer in the boxes below.

| | | | | | | |
|-----|--|--|--|--|--|--|
| Gln | | | | | | |
|-----|--|--|--|--|--|--|

(1)

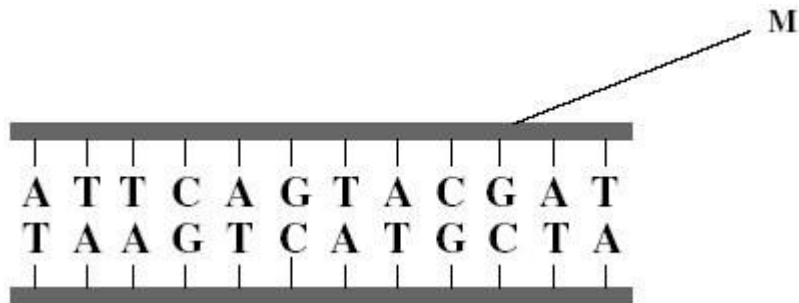
- (e) Explain how a change in a sequence of DNA bases could result in a non-functional enzyme.

(3)

(Total 8 marks)

17

The diagram shows part of a DNA molecule.



(a) Name the **two** components of the part of the DNA molecule labelled **M**.

1. _____

2. _____

(2)

(b) What is the maximum number of amino acids for which this piece of DNA could code?

(1)

(c) Scientists calculated the percentage of different bases in the DNA from a species of bacterium. They found that 14% of the bases were guanine.

(i) What percentage of the bases in this species of bacterium was cytosine?

Answer _____

(1)

(ii) What percentage of the bases in this species of bacterium was adenine?

Answer _____

(1)

- (d) The scientists found that, _____ in a second species of bacterium, 29% of the bases were guanine.

Explain the difference in the percentage of guanine bases in the two species of bacterium.

(2)

(Total 7 marks)

18

- (a) What is meant by a gene?

(2)

The polymerase chain reaction (PCR) can be used to obtain many copies of a particular gene.

- (b) Explain how the strands of DNA are separated during the PCR.

(2)

- (c) In a particular PCR, two different primers are added to the DNA.

- (i) Why are primers required?

(1)

- (ii) Suggest why two different primers are required.

(1)

- (d) Starting with a single molecule of DNA, the polymerase chain reaction was allowed to go through three complete cycles. How many molecules of DNA would be produced?

Answer _____

(1)

(Total 7 marks)

19

Figure 1 shows part of a sarcomere.

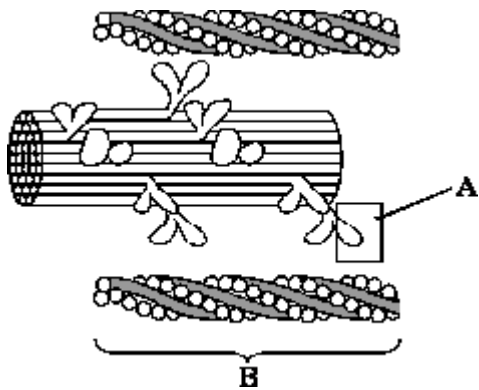


Figure 1

- (a) (i) Name the main protein in structure **B**.

(1)

- (ii) Name the structure in box **A**.

(1)

- (b) (i) Describe how calcium ions cause the myofibril to start contracting.

(2)

(ii) Describe the events that occur within a myofibril which enable it to contract.

(3)

Slow and fast skeletal muscle fibres differ in a number of ways. Slow fibres get their ATP from aerobic respiration while anaerobic respiration provides fast fibres with their ATP. **Figure 2** shows a bundle of fast and slow fibres seen through an optical microscope. The fibres have been stained with a stain that binds to the enzymes which operate in the electron transport chain.

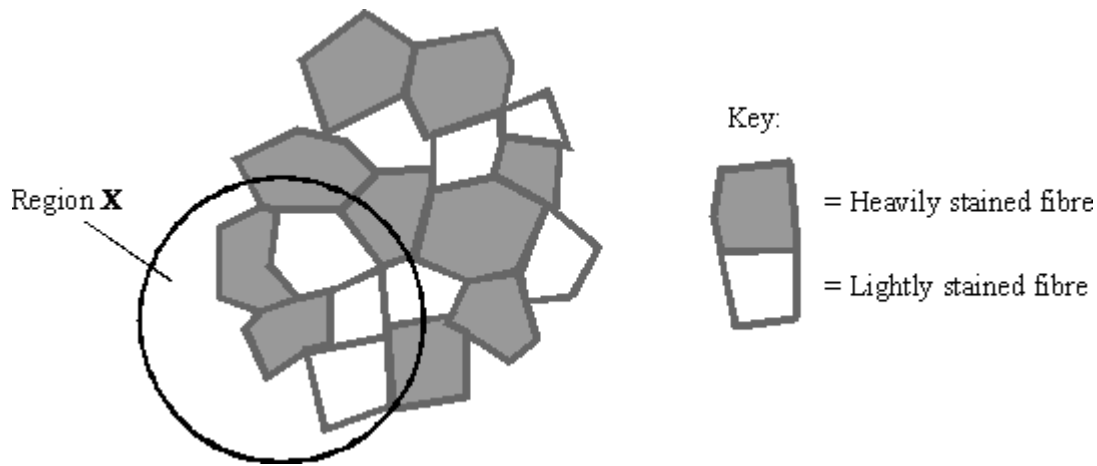


Figure 2

S (c) (i) Describe how you could calculate the percentage of fast fibres in this bundle.

(1)

(ii) The figure calculated by the method in part (c)(i) may not be true for the muscle as a whole. Explain why.

(1)

- (d) The fibres in **Figure 3** correspond to those in region **X** of **Figure 2**. They were stained with a substance that binds to enzymes involved in glycolysis. Shade **Figure 3** to show the appearance of the fibres. Use the shading shown in the key.

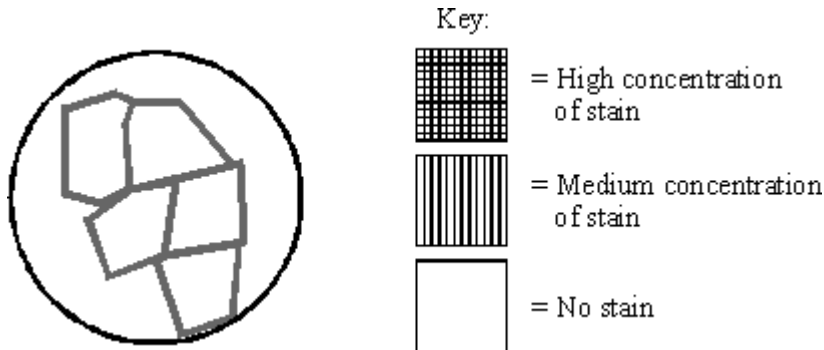


Figure 3

(2)

- S** (e) Recent research has shown that the difference in fibre types is due in part to the presence of different forms of the protein myosin with different molecular shapes.

Explain how a new form of myosin with different properties could have been produced as a result of mutation.

(4)

(Total 15 marks)

20

- (a) (i) What is the role of RNA polymerase in transcription?

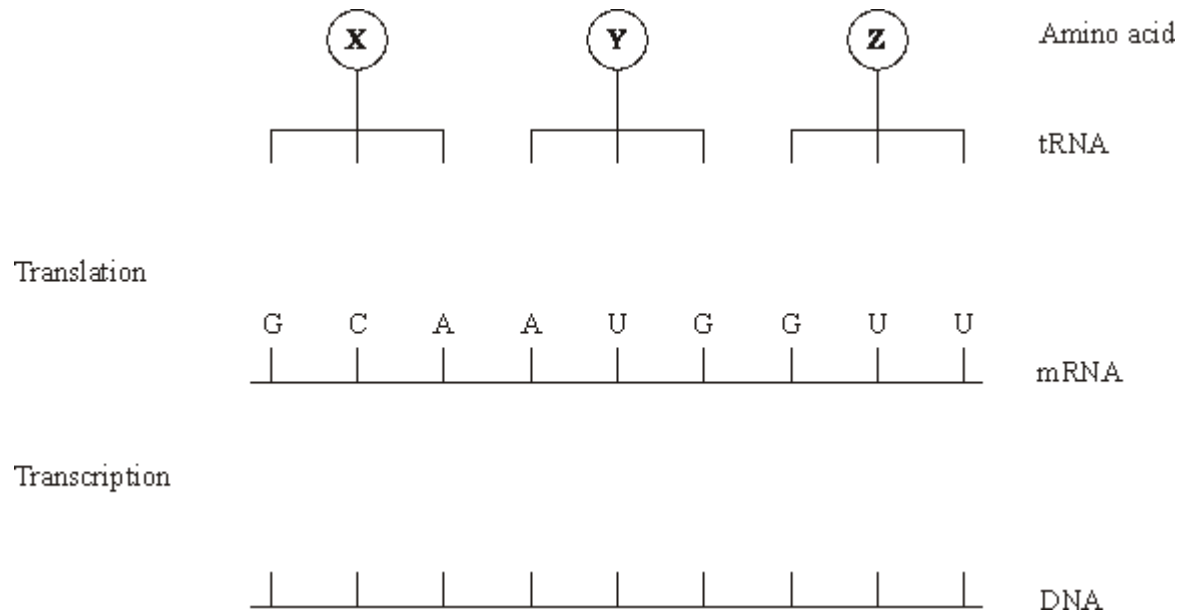
(1)

- (ii) Name the organelle involved in translation.

(1)

(b) **Figure 1** shows some molecules involved in protein synthesis.

Figure 1



Complete **Figure 1** to show

- (i) the bases on the DNA strand from which the mRNA was transcribed;
- (ii) the bases forming the anticodons of the tRNA molecules.

(2)

Figure 2 shows the effects of two different mutations of the DNA on the base sequence of the mRNA. The table shows the mRNA codons for three amino acids.

Figure 2

| | | | | |
|---------------|-------------------|--|--|--|
| | G C A A U G G U U | | | |
| Original mRNA | | | | |
| | G C U A U G G U U | | | |
| Mutation 1 | | | | |
| | G C A A U G G C U | | | |
| Mutation 2 | | | | |

| Amino acid | mRNA codon |
|------------|-------------------|
| methionine | AUG |
| valine | GUC GUU |
| alanine | GCA GCC GCU |

(c) Name the type of mutation represented by mutation 1.

(1)

(d) Use the information in _____ the table to

(i) identify amino acid **X** in **Figure 1**;

(1)

(ii) explain how each mutation may affect the polypeptide for which this section of DNA is part of the code.

Mutation 1 _____

(2)

Mutation 2 _____

(2)

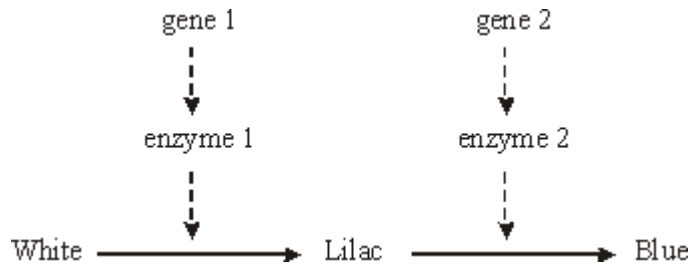
(Total 10 marks)

21

(a) Name **one** mutagenic agent.

(1)

(b) In flax plants the flowers are white, lilac or blue. The diagram shows the pathway by which the flower cells produce coloured pigments.







(i) A deletion mutation occurs in gene 1. Describe how a deletion mutation alters the structure of a gene.

(2)

(ii) Describe and explain how the altered gene could result in flax plants with white-coloured flowers.

(4)

- (iii) Electrophoresis was used to separate the enzymes involved in this pathway. When extracts of the differently coloured flax petals were analysed, four different patterns of bands were produced. In the table, only bands that contain functional enzymes are shown.

| Result of electrophoresis | Colour of petal |
|---|-----------------|
|  | White |
|  | |
|  | |
|  | |

Complete the table to give the colour of the petal from which each extract was taken.

(2)

(Total 9 marks)

22

Mitochondria contain the genes needed for the synthesis of the enzymes involved in the electron transport chain. One of these enzymes is cytochrome oxidase. If a mutation occurs during replication of the mitochondrial genes, functional cytochrome oxidase may not be produced.

- S** Explain why mutation of a mitochondrial gene might result in no functional cytochrome oxidase being produced.

(Total 5 marks)

23

This question should be answered in continuous prose. Quality of Written Communication will be assessed in the answer.

- (i) Starting with mRNA, describe how the process of translation leads to the production of a polypeptide.

(4)

- (ii) Normal tomato plants have an enzyme that softens tomatoes as they ripen. Genetically engineered tomatoes ripen and soften more slowly. A gene was inserted which reduces the amount of softening enzyme produced.

The diagram shows matching parts of the base sequences for the mRNA produced by the gene for the softening enzyme and that produced by the inserted gene.

Softening gene mRNA ...AAUCGGAAU...

Inserted gene mRNA ...UUAGCCUUA...

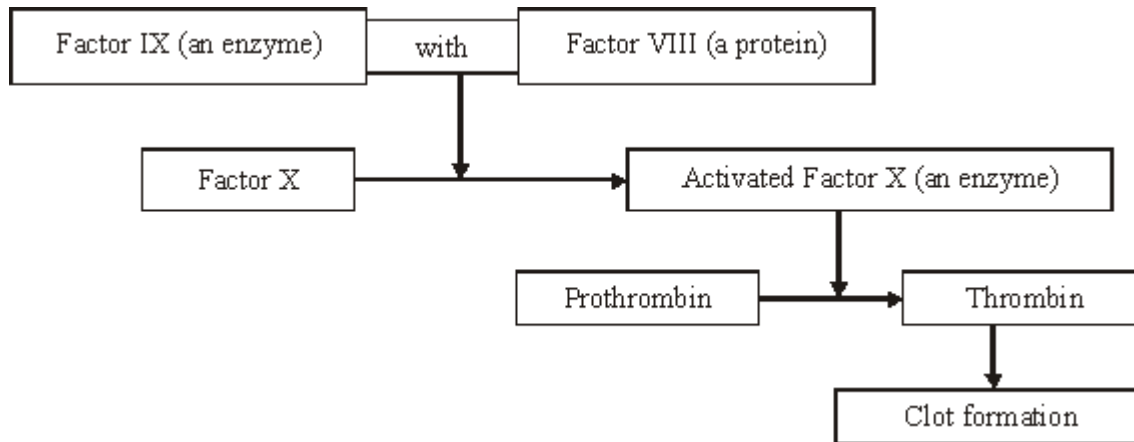
Suggest how the inserted gene reduces the production of the softening enzyme.

(2)

(Total 6 marks)

24

The diagram shows part of the metabolic pathway involved in the clotting of blood in response to an injury.



Haemophilia is a condition in which blood fails to clot. This is usually because of a mutant allele of the gene for Factor VIII.

(a) Explain how mutation could lead to faulty Factor VIII.

(2)

(b) Use information in the diagram to explain how faulty Factor VIII causes haemophilia.

(2)

(c) A boy had haemophilia caused by faulty Factor IX. When his blood was mixed with blood from a haemophiliac with faulty Factor VIII, the mixture clotted. Suggest an explanation for clotting of the mixture.

(2)

(Total 6 marks)

Mark schemes

- 1**
- (a) 1. DNA of eukaryotic cell has non-coding regions / introns within gene
Allow converse: (But) a prokaryotic cell does not have non-coding regions / introns in DNA;
- OR**
- pre-mRNA contains non-coding regions / introns;
2. (After transcription / during modification) these regions are removed from (pre-)mRNA;
Ignore references to 'cells need / bacteria do not need'
- 2
- (b) 1. mRNA longer
- OR**
- Has more nucleotides than tRNA;
2. mRNA is a straight molecule but tRNA is a folded molecule / clover-leaf shaped molecule;
3. mRNA contains no paired bases / hydrogen bonds but tRNA has some paired bases / hydrogen bonds.
- 2 max
- [4]**
- 2**
- (a) Translation.
- 1
- (b) Transfer RNA / tRNA.
- 1
- (c) TAC;
 UAC.
- 2
- (d) Have different R group.
Accept in diagram
- 1
- (e) 1. Substitution would result in CCA / CCC / CCU;
 2. (All) code for same amino acid / proline;
 3. Deletion would cause frame shift / change in all following codons / change next codon from UAC to ACC.
- 3
- [8]**

3

- (a)
1. Helicase;
 2. Breaks hydrogen bonds;
 3. Only one DNA strand acts as template;
 4. RNA nucleotides attracted to exposed bases;
 5. (Attraction) according to base pairing rule;
 6. RNA polymerase joins (RNA) nucleotides together;
 7. Pre-mRNA spliced to remove introns.

6 max

- (b)
1. Polymer of amino acids;
 2. Joined by peptide bonds;
 3. Formed by condensation;
 4. Primary structure is order of amino acids;
 5. Secondary structure is folding of polypeptide chain due to hydrogen bonding;
Accept alpha helix / pleated sheet
 6. Tertiary structure is 3-D folding due to hydrogen bonding and ionic / disulfide bonds;
 7. Quaternary structure is two or more polypeptide chains.

4.

- (c)
1. Hydrolysis of peptide bonds;
 2. Endopeptidases break polypeptides into smaller peptide chains;
 3. Exopeptidases remove terminal amino acids;
 4. Dipeptidases hydrolyse / break down dipeptides into amino acids.

4

- (a)
1. Reduction in ATP production by aerobic respiration;
 2. Less force generated because fewer actin and myosin interactions in muscle;
 3. Fatigue caused by lactate from anaerobic respiration.

- (b) Couple **A**,
1. Mutation in mitochondrial DNA / DNA of mitochondrion affected;
 2. All children got affected mitochondria from mother;
 3. (Probably mutation) during formation of mother's ovary / eggs;

Couple **B**,

- ed;
5. Parents heterozygous;
 6. Expect 1 in 4 homozygous affected.
- 5 max**
- (c)
1. Change to tRNA leads to wrong amino acid being incorporated into protein;
 2. Tertiary structure (of protein) changed;
 3. Protein required for oxidative phosphorylation / the Krebs cycle, so less / no ATP made.
- 4**
- (d)
1. Mitochondria / aerobic respiration not producing much / any ATP;
 2. (With MD) increased use of ATP supplied by increase in anaerobic respiration;
 3. More lactate produced and leaves muscle by (facilitated) diffusion.
- [15]**
- 3**
- 4 max**
- 3**
- 3**

- (e) 1. Enough DNA using PCR;
 2. Compare DNA sequence with 'normal' DNA.

2

[15]

5

- (a) (i) (In all organisms / DNA,) the same triplet codes for the same amino acid;
Accept codon / same three bases / nucleotides
Accept plurals if both triplets and amino acids
Reject triplets code for an amino acid
Reject reference to producing amino acid

1

- (ii) 64;

1

- (b) Splicing;

Ignore deletion references

Accept RNA splicing

1

- (c) (i) 1. (Mutation) changes triplets / codons after that point / causes frame shift;
Accept changes splicing site
Ignore changes in sequence of nucleotides / bases
2. Changes amino acid sequence (after this) / codes for different amino acids (after this);
Accept changes primary structure
Reject changes amino acid formed / one amino acid changed
3. Affects hydrogen / ionic / sulfur bond (not peptide bond);
4. Changes tertiary structure of protein (so non-functional);
Neutral 3-D structure

3 max

- (ii) 1. Intron non-coding (DNA) / only exons coding;
*Context is the **intron***
Do not mix and match from alternatives
Neutral references to introns removed during splicing
1. and 2. Ignore ref. to code degenerate and get same / different amino acid in sequence

2. (So) not translated / no change in mRNA produced / no effect (on protein) / no effect on amino acid sequence;
Accept does not code for amino acids

OR

3. Prevents / changes splicing;
4. (So) faulty mRNA formed;
Accept exons not joined together / introns not removed
5. Get different amino acid sequence;

2 max

[8]

- 6** (a) Locus;
Accept: loci 1
- (b) Differences in DNA / differences in base sequence of DNA;
Accept: number of different alleles / size/variation in gene pool
Reject: genes 1
- (c) 1. Jack Russell (genetic) diversity is (significantly) greatest;
2. Bull terrier (genetic) diversity is (significantly) smallest / is most inbred;
3. Miniature terrier and Airedale terriers are similar;
1-3: do not credit just a list of values
4. Standard deviations do not overlap / do overlap with correct ref to significance;
Reference to significance must be relevant to examples given Max 3
- (d) 1. (Bull terrier) breeding has included a genetic bottleneck/ small population/more inbreeding/ greater selection (pressure);
Accept: founder effect
2. Reduced number of different alleles/size of gene pool;
Reject: decrease in number of genes
Ignore ref to mutations
- OR
3. Miniature (terrier) breeding has included more outbreeding/less selection (pressure);
4. Increased number of different alleles/larger gene pool/more variety of alleles;
Reject if genes used instead of alleles
Reject: lower frequency of alleles
Ignore ref to mutations 2
- [7]
- 7** (a) 250 000; 1

- (b) (i) Loss of 3 bases / triplet = 2 marks;;
'Stop codon / code formed' = 1 mark max unless related to the last amino acid

Loss of base(s) = 1 mark;
eg triplet for last amino acid is changed to a stop codon / code = 2 marks
3 bases / triplet forms an intron = 2 marks
Accept: descriptions for 'intron' eg non-coding DNA
'Loss of codon' = 2 marks

2

- (ii) 1. Change in tertiary structure / active site;
Neutral: change in 3D shape / structure
2. (So) faulty / non-functional protein / enzyme;
Accept: reference to examples of loss of function eg fewer E-S complexes formed

2

[5]

8

- (a) One / an amino acid (can be) coded for by more than one triplet;
Accept codon for triplet
Accept description of triplet – three bases / nucleotides

1

- (b) 1. Triplet / three bases on mRNA;
1. Accept nucleotide for base
1. Accept DNA for mRNA
1. Ignore references to RNA unqualified
2. That code for an amino acid;
2. Accept code for stop / start

2

- (c) (i) To join nucleotides together to form mRNA / premRNA / RNA;
Reject forming base pairs
Accept checking and correcting mismatched base pairs

1

- (ii) Reverse transcriptase;
If they give two enzymes, no mark

1

- (d) GGATCC same as CCTAGG in opposite direction;
Accept reads same both ways / same forward and back
Neutral bases are the opposite of each other / reference to base pairs

1

[6]

- 9** (a) (i) 4; 1
- (ii) 1. Change in amino acid / (sequence of) amino acids / primary structure;
1. Reject = different amino acids are 'formed'
2. Change in hydrogen / ionic / disulphide bonds alters tertiary structure / active site (of enzyme);
2. Alters 3D structure on its own is not enough for this marking point.
3. Substrate not complementary / cannot bind (to enzyme / active site) / no enzyme- substrate complexes form; 3
- (b) 1. Lack of skin pigment / pale / light skin / albino;
2. Lack of coordination / muscles action affected; 2 max
- (c) Founder effect / colonies split off / migration / interbreeding;
Allow description of interbreeding e.g. reproduction between individuals from different populations 1
- [7]**
- 10** (a) (i) 9;
Accept: nine 1
- (ii) Introns / non-coding DNA / junk DNA;
 Start / stop code / triplet;
Neutral: Repeats.
Accept: 'Introns and exons present'.
Reject: 'Due to exons'. 1 max
- (b) Change in amino acid / s / primary structure;
 Change in hydrogen / ionic / disulfide bonds;
 Alters tertiary structure;
Reject: 'Different amino acid is formed' – negates first marking point.
Neutral: Reference to active site. 3

(c) Number of bases

| | Number of bases | | | |
|----------|-----------------|-----------|-----------|-----------|
| | C | G | A | T |
| Strand A | 26 | 19 | 20 | 9 |
| Strand B | 19 | 26 | 9 | 20 |

Second column correct;

Columns three and four correct;

2

[7]

11

(a) (i) Phosphate and ribose;

Accept in either order. Both correct for one mark.

For phosphate accept PO_4 / Pi / \textcircled{P} but not P.

Do not accept phosphorus.

Ignore references to pentose / sugar.

1

(ii) TAGGCA;

1

(b) (i) Does not contain hydrogen bonds / base pairs / contains codons / does not contain anticodon / straight / not folded / no amino acid binding site / longer;

Assume that "it" refers to mRNA.

Do not accept double stranded.

1

(ii) (pre-mRNA) contains introns / mRNA contains only exons;

Assume that "it" refers to pre-mRNA.

Accept non-coding as equivalent to intron.

1

(c) (i)

| Part of chromosome | U |
|--------------------|----|
| Middle | 18 |
| End | 21 |

One mark for both figures correct

1

- (ii) 1. Have different (base) sequences / combinations of (bases);
 2. (Pre-mRNA) transcribed from different DNA / codes for different proteins;
- 2
[7]

12

- (a) More than one polypeptide / chain;
Ignore references to haem / other groups
- 1
- (b) (i) 141;
- 1
- (ii) 1. Stop / start sequences;
 2. Non coding DNA (in the gene) / introns / multiple repeats / junk DNA;
Do not credit "some bases repeated"
 3. Two chains / a non-coding strand / complementary base pairs;
 4. Addition of base by mutation;
- 2 max
- (c) Different primary structure / amino acids / different number of polypeptide chains;
Question is about haemoglobin so do not credit differences in DNA
- 1
- (d) 1. Low partial pressure of oxygen in lungs;
 2. (Llama) haemoglobin able to load more oxygen / (llama) haemoglobin saturated (at low / particular partial pressure of oxygen);
 3. Higher affinity for oxygen;
The terms used in the graph (or near approximations) should be used in this answer.
Ignore references to unloading
The answer must relate to llamas
- 3
[8]

13

- (a) Banding pattern changes as cheetah gets older / difficult to judge as tail is short / fluffy;
- 1
- (b) (i) Mean not (always) a whole number;
 Standard deviation not (always) zero;
- 2
- (ii) Movement of tail / angle of sight / confused it with another band / subjective estimation;
*Accept reference to **Figure 1***
E.g. Bands 2 and 3 have same thickness but look different
- 1

- (c) Band width not the same on both sides of tail; 1
- (d) Offspring of the same family will be more similar genetically;
As have same mother (and father) / parent;
Expect to see more differences in randomly chosen cheetahs; 3

[8]

14

- (a) Introns; 1
- (b) Ile Gly Val Ser; 1
- (c) (i) Has no effect / same amino acid (sequence) / same primary structure;
Q Reject same amino acid formed or produced. 1
- Glycine named as same amino acid; 1
- It still codes for glycine = two marks.*
- (ii) Leu replaces Val / change in amino acid (sequence) / primary structure;
Change in hydrogen / ionic bonds which alters tertiary structure / active site;
Q Different amino acid formed or produced negates first marking point.
- Substrate cannot bind / no longer complementary / no enzyme-substrate complexes form;
Active site changed must be clear for third marking point but does not need reference to shape. 3
- (d) (i) Interphase / S / synthesis (phase); 1
- (ii) DNA / gene replication / synthesis occurs / longest stage;
Allow 'genetic information' = DNA.
Allow 'copied' or 'formed' = replication / synthesis 1

[9]

15

(a)

| | | |
|------|---|---|
| DNA | ✓ | 2 |
| mRNA | ✗ | 1 |
| tRNA | ✓ | 1 |

*One mark for each correct column
 Regard blank as incorrect in the context of this question
 Accept numbers written out: two, one, one*

2

(b) (i) Marking principles

1 mark for complete piece transcribed;

*Correct answer
 UGU CAU GAA UGC UAG*

1 mark for complementary bases from sequence transcribed;

*but allow 1 mark for complementary bases from section transcribed,
 providing all four bases are involved*

2

(ii) Marking principle

1 mark for bases corresponding to exons taken from (b)(i)

*Correct answer
 UGU UGC UAG
 If sequence is incorrect in (b)(i), award mark if section is from
 exons. Ignore gaps.*

1

[5]

16

(a) (i) Deoxyribose;

pentose / 5C sugar = neutral

1

(ii) Phosphate / Phosphoric acid;

phosphorus / P = neutral

1

(b) Hydrogen (bonds);

1

(c) 381 / 384 / 387;

1

(d) (Gln) Met Met Arg Arg Arg Asn;

1

- (e) Change in (sequence of) amino acids / primary structure;
 Change in hydrogen / ionic / disulfide bonds leads to change in tertiary structure / active site (of enzyme);

Substrate cannot bind / no enzyme-substrate complexes form;

Q Reject = different amino acids are formed

3

[8]

17

- (a) Phosphate;

Deoxyribose;

Q Candidates must specify deoxyribose. This term is a specification requirement.

Ignore anything that is not incorrect.

2

- (b) 4;

1

- (c) (i) 14;

1

- (ii) 36;

If (c)(i) incorrect accept [50 – (c)(i)]

1

- (d) Different genes;

Different (DNA) base sequences;

2

[7]

18

- (a) a length of DNA;
 that codes for a single protein / polypeptide;

2

- (b) by heating;
 to break the H-bonds (between complementary bases);

2

- (c) (i) to allow the DNA polymerase to attach / start addition of nucleotides / mark start and end of sequence to be copied / prevents strands re-joining;

1

- (ii) because the sequences at the ends of the target sequence are different / one is at the beginning and one at the end;

1

- (d) 8;

accept 7

1

[7]

- 19**
- (a) (i) actin (*Accept* tropomyosin); 1
- (ii) myosin head; 1
- (b) (i) Ca²⁺ binds to [part of] the actin / troponin;
this causes tropomyosin to be displaced;
uncovers [myosin] binding sites [on actin] / allows actin to bind; max 2
- (ii) myosin heads bind to actin / cross bridge formation /
actomyosin formed;
myosin heads / crossbridges swivel / ratchet mechanism;
causing actin to slide relative to myosin;
energy provided by hydrolysis of ATP; max 3
- (c) (i) (number lightly stained fibres / total number of fibres) × 100;
(actual numbers are 10 / 18 × 100) 1
- (ii) sample not representative / large enough / individual muscle fibres
different sizes / contain different number of myofibrils; 1
- (d) all some stain = 1
fast dark and slow lighter = 2 2
- (e) change in base sequence in DNA / addition / deletion / substitution of a base in DNA
of the gene which codes for myosin;
change in amino acid sequence / primary structure;
causes a different tertiary structure;
which alters the binding properties of myosin; 4
- [15]**
- 20**
- (a) (i) join / attach nucleotides, to form a strand / along backbone / phosphodiester bonds;
(*reject reference to H bonds, complementary base pairing*) 1
- (ii) ribosome / RER; 1
- (b) (i) CGTTACCAA; 1
- (ii) CGU UAC CAA; 1
- (c) substitution; 1

- (d) (i) alanine; 1
- (ii) (mutation 1)
 no change(to sequence of amino acids);
 codon for alanine / degenerate codon / same amino acid coded for; 2
- (mutation 2)
 (change in sequence) valine replaced by alanine / codon for alanine;
 folding / shape / tertiary structure / position of bonds may change;
(reject peptide bonds) 2

[10]

21

- (a) high energy radiation / ionising particles;
 named particles / α , β , γ ;
 colchicine;
 x rays / cosmic rays;
 uv (light);
 carcinogen / named carcinogen;
 mustard gas / phenols / tar (qualified); 1 max
- (b) (i) removal of one or more bases / nucleotide;
 frameshift / (from point of mutation) base sequence change; 2
- (ii) sequence of bases in mRNA would change;
 (sequence of) amino acids different / different primary structure;
 (active site / enzyme 1) changed tertiary shape / changed active sites;
white pigment does not bind;
 lilac pigment not produced / white pigment remains unchanged /
 enzyme 1 does not function; 4 max
- (iii) blue and lilac; white;

| |
|------------------------|
| <i>colour of petal</i> |
| <i>(white)</i> |
| blue |
| lilac; |
| white; |

2

[9]

22 change in base / nucleotide (in DNA);
 change in base sequence of mRNA / change in codons / idea of frameshift following deletion or addition / incorrect tRNA / anticodon;
 incorrect amino acids / different primary structure / formation of new stop codon;
 different tertiary structure / different 3D structure / different polypeptide / shortened polypeptide;
 different shape of active site / no active site present;

[5]

23 (i) mRNA attaches to ribosome;
 codon on mRNA;
 binds to an anti-codon on tRNA;
 each tRNA brings a specific amino acid;
 sequence of codons / bases on mRNA determines order of amino acids;
 formation of peptide bonds / amino acids joined by condensation reactions;

4 max

(iii) inserted gene / mRNA complementary to normal gene / mRNA;
 binds to it to prevent protein synthesis / form double strand / prevents mRNA binding to ribosomes;
 will not stop all translation, some mRNA reaches ribosomes / because not all mRNA is bound by inserted gene mRNA;

2 max

[6]

24 (a) mutation changes the amino acid sequence / primary structure of Factor VIII protein;
 changes the tertiary structure / 3D shape;

2

(b) (mutant) Factor VIII protein is non-functional / does not work with Factor IX;
 so no conversion of Factor X to active form and pathway blocked;

2

(c) boy's blood contains (active) Factor VIII;
 Factor VIII haemophiliac's blood contains (active) Factor IX;
 the mixture has both Factors and so the pathway can complete / blood clots;

2 max

[6]