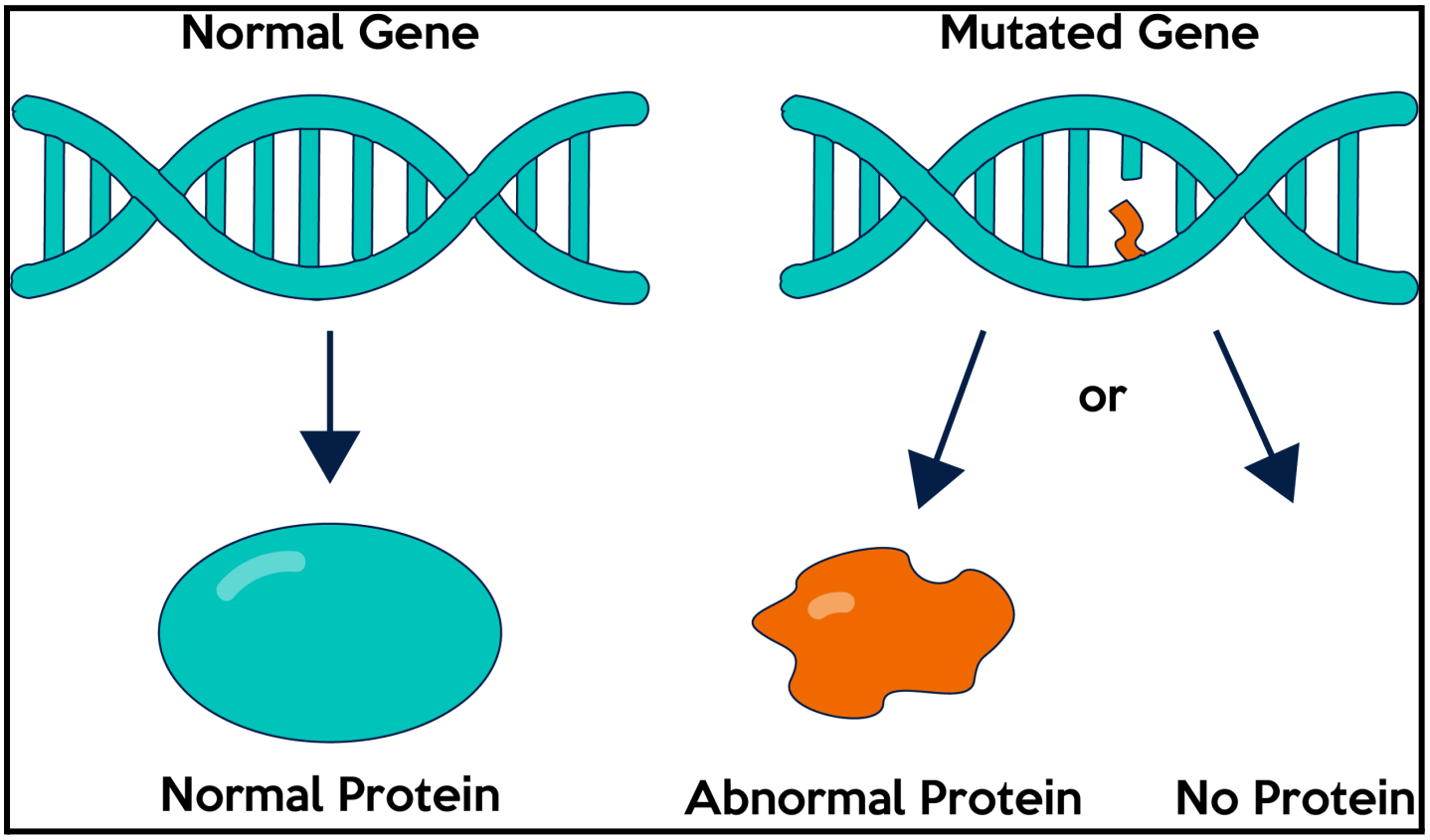
# A Level Biology

## DNA - Mutation.

## Name:



**Retrieval Practice.**

1. **How are DNA & RNA best described?**
2. **List three key differences between DNA & RNA.**
3. **How is DNA copied?**
4. **What is the triplet code?**
5. **How many codons can be produced from the triplet code?**
6. **Which enzyme is responsible for transcription and where does transcription occur?**
7. **What is splicing?**
8. **Where does splicing occur?**
9. **What is an anticodon and where would it be found?**
10. **Compare mRNA & tRNA**
11. **How and where does translation occur?**

**Mutation**

- **Understand the term gene mutation as illustrated by base deletions, insertions and substitutions.**
- **Understand the effect of point mutations on amino acid sequences, as illustrated by sickle cell anaemia in humans.**

RNA codon table

| 1st position | 2nd position U | 2nd position C | 2nd position A | 2nd position G | 3rd position |
|---|---|---|---|---|---|
| U | Phe | Ser | Tyr | Cys | U |
| | Phe | Ser | Tyr | Cys | C |
| | Leu | Ser | stop | stop | A |
| | Leu | Ser | stop | Trp | G |
| C | Leu | Pro | His | Arg | U |
| | Leu | Pro | His | Arg | C |
| | Leu | Pro | Gln | Arg | A |
| | Leu | Pro | Gln | Arg | G |
| A | Ile | Thr | Asn | Ser | U |
| | Ile | Thr | Asn | Ser | C |
| | Ile | Thr | Lys | Arg | A |
| | Met | Thr | Lys | Arg | G |
| G | Val | Ala | Asp | Gly | U |
| | Val | Ala | Asp | Gly | C |
| | Val | Ala | Glu | Gly | A |
| | Val | Ala | Glu | Gly | G |

Amino Acids

Ala: Alanine
Arg: Arginine
Asn: Asparagine
Asp:Aspartic acid
Cys:Cysteine
Gln: Glutamine
Glu: Glutamic acid
Gly: Glycine
His: Histidine
Ile: Isoleucine
Leu: Leucine
Lys: Lysine
Met: Methionine
Phe: Phenylalanine
Pro: Proline
Ser: Serine
Thr: Threonine
Trp: Tryptophane
Tyr: Tyrosisne
Val: Valine

**No mutation - Normal DNA.**

**Original DNA Sequence**

| | | | | | |
|---|---|---|---|---|---|
| TAC | GTG | ACA | CGG | CAT | ATT |

**mRNA Sequence**

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**Polypeptide amino acid sequence.**

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**Consequence**

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**Substitution type 1 - Silent Mutation.**

**Original DNA Sequence**

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|---|---|---|---|---|---|
| TAC | GTG | ACA | CGG | CAT | ATT |

**Mutated DNA Sequence.**

| | | | | | |
|---|---|---|---|---|---|
| TAC | GTA | ACA | CGG | CAT | ATT |

**mRNA Sequence**

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**Amino acid sequence.**

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**Consequence**

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**Substitution type 2 - missense.**

**Original DNA Sequence.**

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|---|---|---|---|---|---|
| TAC | GTG | ACA | CGG | CAT | ATT |

**Mutated DNA Sequence.**

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|---|---|---|---|---|---|
| TAC | GTT | ACA | CGG | CAT | ATT |

**mRNA Sequence**

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**Polypeptide amino acid sequence.**

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**Consequence**

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**Substitution Type 3 - Non Sense mutation.**

**Original DNA Sequence.**

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| TAC | GTG | ACA | CGG | CAT | ATT |

**DNA Sequence**

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|---|---|---|---|---|---|
| TAC | GTG | ACT | CGG | CAT | ATT |

**mRNA Sequence**

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**Polypeptide amino acid sequence.**

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**Consequence**

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**Deletion mutation (causing frameshift)**

**Original DNA Sequence.**

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|---|---|---|---|---|---|
| TAC | GTG | ACA | CGG | CAT | ATT |

**DNA Sequence**

| | | | | | |
|---|---|---|---|---|---|
| TAC | GTG | ACA | GGC | ATA | TTT |

**mRNA Sequence**

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**Polypeptide amino acid sequence.**

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**Consequence**

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**insertion mutation (causing frameshift)**

**Original DNA Sequence.**

| | | | | | |
|---|---|---|---|---|---|
| TAC | GTG | ACA | CGG | CAT | ATT |

**DNA Sequence**

| | | | | | | |
|---|---|---|---|---|---|---|
| TAC | TGT | GAC | ACG | GCA | TAT | TTT |

**mRNA Sequence**

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|---|---|---|---|---|---|---|
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**Polypeptide amino acid sequence.**

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**Consequence**

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**Sickle cell Anemia**

**Wild Type (sense) DNA**

| ATG | GTG | CAC | CTG | ACT | CCT | GAG | AAG | TCT | GCC |
|---|---|---|---|---|---|---|---|---|---|

**Wild Type (antisense) DNA**

| | | | | | | | | | |
|---|---|---|---|---|---|---|---|---|---|

**mRNA Sequence**

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|---|---|---|---|---|---|---|---|---|---|

**Amino acid sequence**

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**Sickle cell mutation (sense strand)**

| ATG | GTG | CAC | CTG | ACT | CCT | GTG | AAG | TCT | GCC |
|---|---|---|---|---|---|---|---|---|---|

**Sickle Cell (antisense Strand)**

| | | | | | | | | | |
|---|---|---|---|---|---|---|---|---|---|

**mRNA Sequence**

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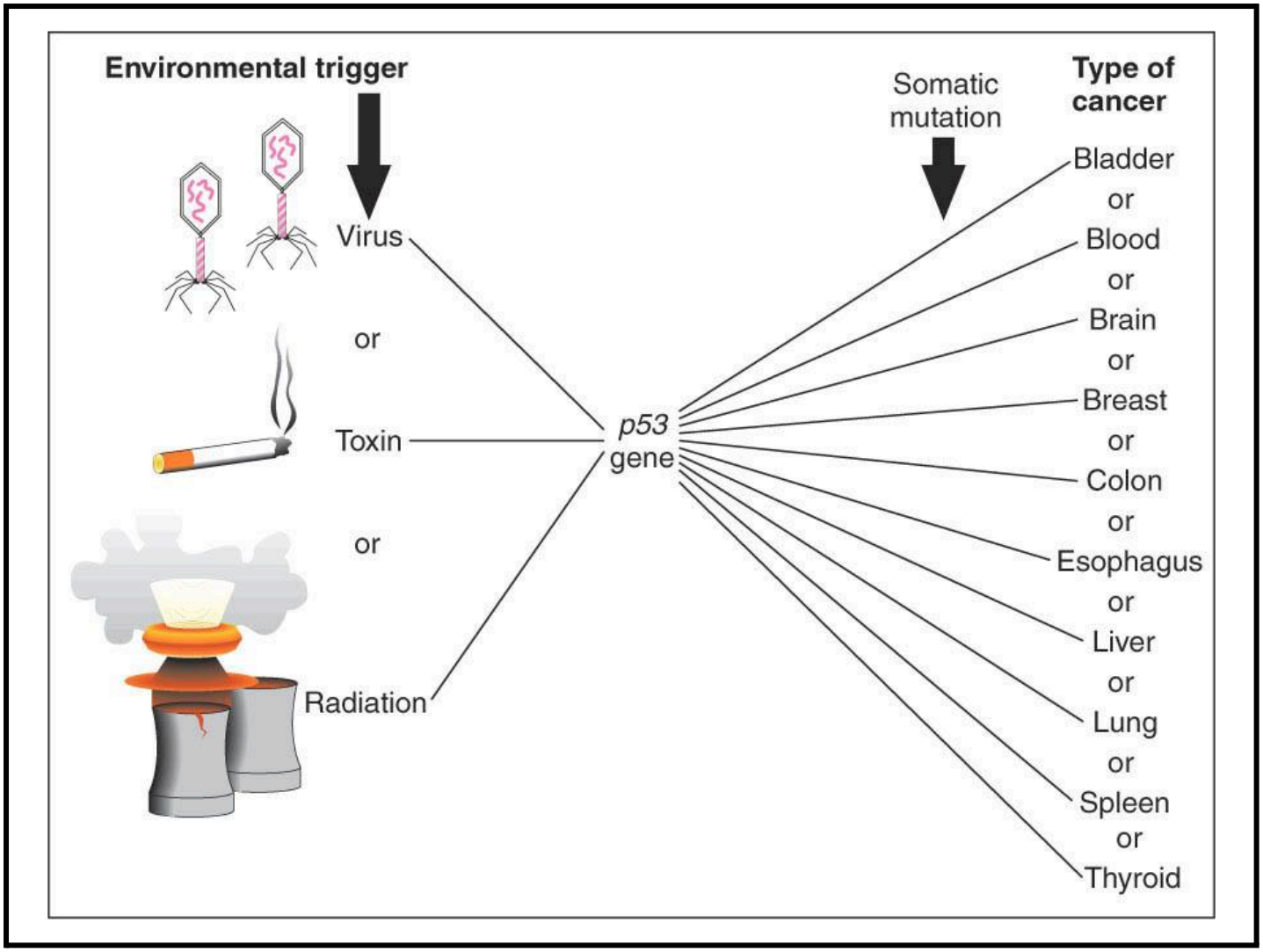
|---|---|---|---|---|---|---|---|---|---|

**Amino acid sequence**

| | | | | | | | | | | |
|---|---|---|---|---|---|---|---|---|---|---|

**Consequence**

**What causes mutations?**



Examination Practice

Q1.

(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

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|---|
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(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)**

**Q2. Lysozyme is an enzyme consisting of a single polypeptide chain of 129 amino acids.**

**(a) What is the minimum number of nucleotide bases needed to code for this enzyme?**

| |
|---|
| |

**(1)**

**(b) The diagram shows the sequence of bases in a section of the mRNA strand used to synthesise this enzyme.**

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

**(i) Give the DNA sequence which would be complementary to the first four bases in this section of mRNA.**

| |
|---|
| |

**(1)**

**(ii) How many different types of tRNA molecule would attach to the section of mRNA shown in the diagram?**

| |
|---|
| |

**(1)**

**(c) Give two factors which might increase the frequency at which a mutation in DNA occurs.**

| |
|---|
| **1.** |
| **2.** |

**(2)**

**(d) Two single base mutations occurred in the DNA coding for this section of mRNA. These mutations caused an alteration in the sequence of amino acids in the enzyme. The diagram shows the original and altered sequences of amino acids.**

| | | | | | | | |
|---|---|---|---|---|---|---|---|
| **Original amino acid sequence** | **Gly** | **Leu** | **Ser** | **Tyr** | **Gly** | **Arg** | **Tyr** |
| **Original mRNA base sequence** | **GGU** | **CUU** | **UCU** | **UAU** | **GGU** | **AGA** | **UAU** |

| | | | | | | | |
|---|---|---|---|---|---|---|---|
| **Altered amino acid sequence** | **Gly** | **Leu** | **Tyr** | **Leu** | **Trp** | **Arg** | **Tyr** |
| **Altered mRNA base sequence** | **GGU** | **CUU** | | | | **AGA** | **UAU** |

**(i) Use the mRNA codons provided in the table to complete the altered mRNA base sequence in the diagram.**

| Amino acid | mRNA codons which can be used |
|---|---|
| Arg | AGA |
| Gly | GGU |
| Leu | CUU or UUA |
| Ser | UCU |
| Trp | UGG |
| Tyr | UAU or UAC |

**(1)**

**(ii) Use the information provided to determine the precise nature of the two single base mutations in the DNA.**

**(3)**

**(Total 9 marks)**

**Q3.(a)** **A mutation can lead to the production of a non-functional enzyme. Explain how.**

**(6)**

**Q5.(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?**

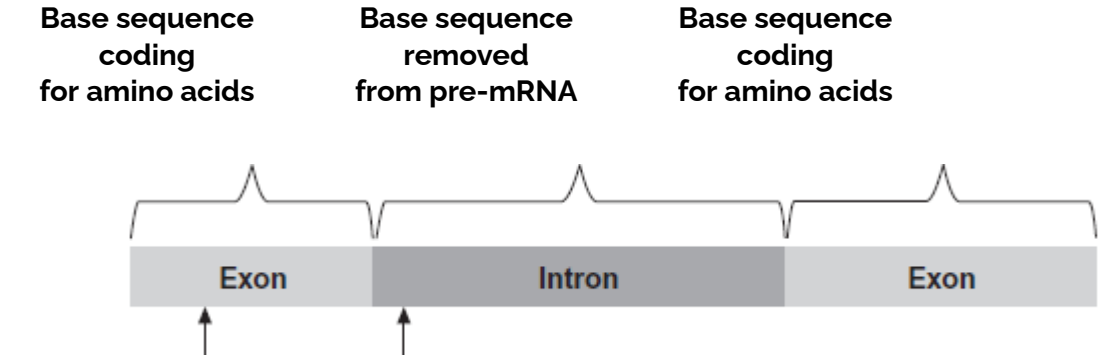
**(1)**

**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.**

(3)

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

**Explain your answer.**

(2)

**(Total 8 marks)**

**Q7.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 |

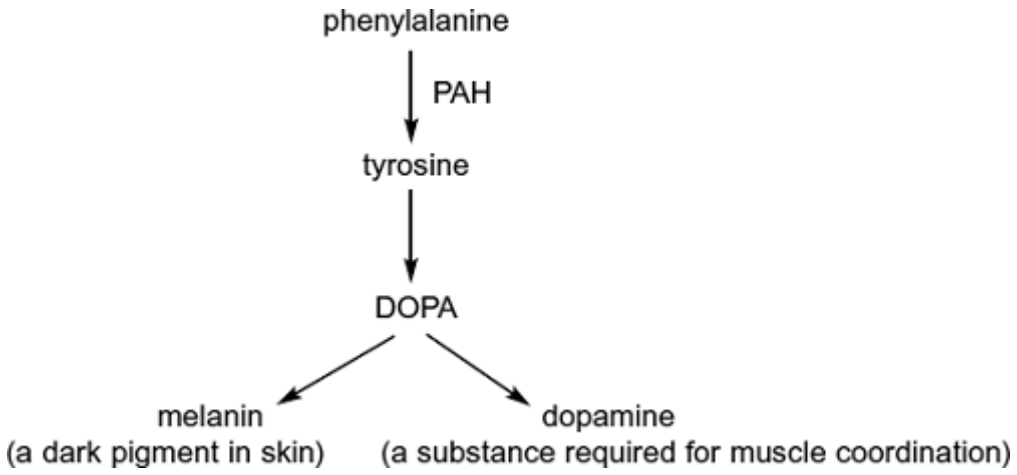
**(a) (i) What is the maximum number of amino acids for which this base sequence could code?**

(1)

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

(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)**

**M1.**

**(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]**

**M2. (a) 387;**

**1**

**(b) (i) CCAG;**

**1**

**(ii) 5;**

**1**

**(c)** **high energy radiation / X rays / ultraviolet light / gamma rays;**
**high energy particles / alpha particles / beta particles;**
**named chemical mutagens e.g. benzene / caffeine / pesticide / mustard gas / tobacco tar / free radicals;**
***(two named examples of any of the above = 2 marks)***
**length of time of exposure (to a mutagen);**
**dosage (of mutagen);**

**2 max**

**(d)** **(i)** **UAC UUA UGG;**

**1**

**(ii)** **addition and deletion (of bases / nucleotides);**
**thymine added;**
**adenine deleted;**
***(addition of thymine and deletion of adenine = 3 marks)***
***(allow addition of adenine (RNA) and deletion of uracil (RNA) = 2 marks)***

**3**

**[9]**

**M3.(a) 1. Change / mutation in base / nucleotide sequence (of DNA / gene);**

***Ignore: references to changing base-pairing***
***Accept: affect for change, if in correct context***
***Accept: changes triplets / codons***

**2. Change in amino acid sequence / primary structure (of enzyme);**
***Accept: different amino acid(s) coded for***
***Q Reject: different amino acids produced / formed / made***

**3. Change in hydrogen / ionic / disulfide bonds;**
***Accept: references to sulfur bonds***

**4. Change in the tertiary structure / shape;**
***Neutral: alters 3D structure / 3D shape***

**5. Change in active site;**

**6. Substrate not complementary / cannot bind (to enzyme / active site) / no enzyme-substrate complexes form.**
***Accept: no E S complexes form***

**6**

**(b) 1. Non-SR strain falls more / SR strain falls less / up to 10(µg / cm−3);**
***Must include 10 but only required once in either MP1 or MP2***
***Ignore: units or absence of***
***This must be a comparative statement***

**2. Above 10(µg / $cm^{-3}$), SR strain levels out / off and non-SR strain continues to decrease;**

**3. Greater difference between strains with increasing concentration of antibiotic.**
***This must be a comparative statement***

**2 max**

**(c) 1. Division stopped (of both strains by scientist);**
***Reject: references to mitosis stopping***

**2. SR strain still more resistant / fewer die / none die (at higher concentrations of antibiotic).**
***Accept: SR strain and non-SR strain would be similar if resistance is due to only stopping division***
***Need some comparison with non-SR***

**2**

(d) 1. Make a competitive / non-competitive inhibitor;

*Mark in pairs*

*either MP1 and MP2 OR MP3 and MP4*

2. Competitive competes with / blocks active site / non-competitive inhibitor affects / changes active site;

*Do not mix and match*

OR

3. (Make a drug) that inhibits / denatures / destroys enzyme / stringent response;

*Accept: drug that 'knocks out' / destroys enzyme*

4. Give at the same time as / before an antibiotic.

**2 max**

(e) (SR strain)

1. Fewer free radicals (than non-SR);

*Note: has to be comparative statement*

2. Produces more catalase (than non-SR);

*Accept converse statements for non-SR.*

3. Catalase (might be) linked to production of fewer free radicals / breaking down / removing free radicals.

*Accept: hydrolysis of radicals by catalase.*

**3**

**[15]**

M4.(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.

**3**

(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,**

4. **Mutation in nuclear gene / DNA in nucleus affected;**
5. **Parents heterozygous;**
6. **Expect 1 in 4 homozygous affected.**

**4 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.**

**3**

**(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.**

**3**

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

**2**

**[15]**

**M5.(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 <u>intron</u>*

*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]

(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]

M7.(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]**