## Mark schemes

(a) 1. Degenerate: more than one (base) triplet for each amino acid;
2. Non-overlapping: each base is part of only one triplet.Accept codon (as would be applicable to mRNA code)
(b) A = adenine
$C=$ cytosine
$G=$ guanine
$\mathrm{U}=$ uracil
All four correct = 2
One error = 1
Two or more errors = 0
2 max
(c) AGT ;
[5] (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 shapedmolecule;
3. mRNA contains no paired bases / hydrogen bonds but tRNA has some paired bases /hydrogen bonds.

$$
2 \max [4]
$$

(a) Translation.
(b) Transfer RNA / tRNA.
(c) TAC;

UAC.
(d) Have different R group.

Accept in diagram
(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 nextcodon from UAC to ACC.
[8] (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,
4. Mutation in nuclear gene / DNA in nucleus affected;
5. Parents heterozygous;
6. Expect 1 in 4 homozygous affected.
(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 / noATP made.
(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.
(e) 1. Enough DNA using PCR;
2. Compare DNA sequence with 'normal' DNA.
(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.
(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.

6

| Feature | Bacterium | Human <br> immunodeficiency virus <br> (HIV) particle |
| :--- | :---: | :---: |
| RNA | $\checkmark$ | $\checkmark$ |
| Cell wall | $\checkmark$ |  |
| Enzyme molecules | $\checkmark$ | $\checkmark$ |
| Capsid |  | $\checkmark$ |

correct vertical column for each
(b) 1. (Complementary) nucleotides/bases pair

OR
$A$ to $T$ and $C$ to $G$;
Ignore '(DNA polymerase) forms base pairs/nucleotide pairs'
2. DNA polymerase;
3. Nucleotides join together (to form new strand)/phosphodiester bonds form;

Ignore '(DNA polymerase) forms base pairs/nucleotide pairs'
If clearly writing rote answer about DNA replication $\underline{2} \max$ e.g.
helicase or separating strands
(c) 1. DNA double stranded/double helix and mRNA single-stranded;

Contrast requires both parts of the statement
2. DNA (very) long and RNA short;

Accept 'RNA shorter' or 'DNA bigger/longer'
3. Thymine/T in DNA and uracil/U in RNA;
4. Deoxyribose in DNA and ribose in RNA; $\boldsymbol{R}$ Deoxyribonucleic/ ribonucleic acid

Ignore ref. to histones
Ignore ref. to helix and straight chain alone
5. DNA has base pairing and mRNA doesn't/ DNA has hydrogen bonding and mRNA doesn't;
6. DNA has introns/non-coding sequences and mRNA doesn't; Ignore ref to splicing

3 max
[8] (a) Quaternary (structure);

Accept phonetic spelling eg quarternary/quarternery $/ 4^{\circ}$
Award no mark for quaternary as part of a list
(b) 423;
(c) 1. Oxyhaemoglobin formed/ haemoglobin is loaded/uptakes/associates/binds with oxygen in area of higher $\mathrm{ppO}_{2}$ / in gas exchange surface/lungs/gills;

Reference to "react with" = max 1
Accept: reversible interaction with oxygen
Ignore: haemoglobin is carried / contained in red blood cells
2. (oxygen) unloaded/dissociates from/released (in area of lower $\mathrm{ppO}_{2}$ / in capillaries/to cells/tissues);

2
(d) (i) $56(\%)$;

Accept responses in the range 54-58(\%)
1
(ii) 1. (Anaemia curve shifted to right) haemoglobin has lower affinity for oxygen / binds less tightly;
Assume reference is to haemoglobin of anaemia unless stated
2. releases more oxygen / oxygen is released quicker / oxygen dissociates/ unloads more readily to muscles/tissues/cells;
3. (For) respiration;

Accept: even with a lower haemoglobin concentration / meet demand for ATP/energy;
(a) 1. (Reaction with ATP) breaks/allows binding of myosin to actin/ actinomyosin bridge;
2. Provides energy to move myosin head;

1. Credit 'breaks' or 'allows' binding to actin (because cyclical)
2. Allow in context of 'power stroke' or 're-cocking' (becausecyclical)
3. Ignore contraction on its own
(b) (i) Any value between 68.5 and 69.49 (\%);;

If get difference of 0.9 but calculation of percentage incorrect, then award 1 mark;
(ii) (Mutant mice)

1. Unable to make phosphocreatine/ less phosphateavailable to make/recycle ATP;
2. So less energy/so less ATP available for contraction/fastmuscle fibres;
1 and 2. Reject production/creation of energy once
2 Accept less energy for grip
3. Accept no energy/no ATP for contraction/fast muscle fibres
(c) 1. (Heterozygous) have one dominant/normal allele (for creatineproduction);
4. (This) leads to production of enough/normal amount ofcreatine;
5. Accept has one allele/one copy of the gene for/that is making creatine
(a) (i) 1. (Tumour suppressor) gene inactivated / not able to control / slow down cell division;

Ignore: references to growth
2. Rate of cell division too fast / out of control.

1 and 2 Accept: mitosis
1 and 2 Reject: meiosis
(ii) 1. (Genetic) code degenerate;

Accept: codon for triplet
Accept description of degenerate code, e.g. another triplet codes for the same amino acid
2. Mutation in intron.

Accept: mutation in non-coding DNA
(b) 1. Antibody has specific tertiary structure / binding site / variable region;Do not accept explanations involving undefined antigen
2. Complementary (shape / fit) to receptor protein / GF / binds to receptor protein /to GF;

Ignore: same shape as receptor protein / GF
3. Prevents GF binding (to receptor).
[6] (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
(b) Splicing;

Ignore deletion references
Accept RNA splicing
(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 aminoacids (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
(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;
(a) 1. Sugar-phosphate (backbone) / double stranded / helix so provides strength / stability
/ protects bases / protects hydrogen bonds;
Must be a direct link / obvious to get the mark
Neutral: reference to histones
2. Long / large molecule so can store lots of information;
3. Helix / coiled so compact;

Accept: can store in a small amount of space for 'compact'
4. Base sequence allows information to be stored / base sequence codes foramino acids / protein;

Accept: base sequence allows transcription
5. Double stranded so replication can occur semi-conservatively / strands can act as templates / complementary base pairing / A-T and G-C so accurate replication / identical copies can be made;
6. (Weak) hydrogen bonds for replication / unzipping / strand separation / many hydrogen bonds so stable / strong;

Accept: 'H-bonds' for 'hydrogen bonds'
(b) 1. (Mutation) in E produces highest risk / 1.78;
2. (Mutation) in D produces next highest risk / 1.45;
3. (Mutation) in C produces least risk / 1.30; Must be stated directly and not implied
$\boldsymbol{E}>\boldsymbol{D}>\boldsymbol{C}=3$ marks
Accept: values of $0.78,0.45$ and 0.30 for MP1, MP2 and MP3 respectively
If no mark is awarded, a principle mark can be given for the idea that all mutant alleles increase the risk
(c) 180;
(d) (Similarities):

1. Same / similar pattern / both decrease, stay the same then increase;
2. Number of cells stays the same for same length of time; Ignore: wrong days stated

## (Differences):

(Per unit volume of blood)
3. Greater / faster decrease in number of healthy cells / more healthy cells killed /healthy cells killed faster;

Accept: converse for cancer cells
Accept: greater percentage decrease in number of cancer cells / greater proportion of cancer cells killed
4. Greater / faster increase in number of healthy cells / more healthy cellsreplaced / divide / healthy cells replaced / divide faster;

Accept: converse for cancer cells
For differences, statements made must be comparative
(e) 1. More / too many healthy cells killed;
2. (So) will take time to replace / increase in number; Neutral: will take time to 'repair'
3. Person may die / have side effects;
[15] (a) 250000 ;
12
(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
(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
[5] (a) 1. (Protein / molecule) that moves from cytoplasm to DNA;

Accept 'it' as TF.
Accept moves into nucleus
2. (TF) binds to specific gene / genes / to specific part of / site on DNA / binds topromoter / RNA polymerase;

Accept regulator / enhancer region
3. Leads to / blocks (pre)mRNA production / allows / blocks binding of RNApolymerase (to DNA) / allows RNA polymerase to work;

Ignore translation unless context wrong
Max 1 if refer to oestrogen as a transcription factor
(b) 1. (Binding to CREB) prevents transcription / mRNA formation;Accept that lack of protein leaves NAD reduced
2. (Binding of huntingtin) prevents production / translation of protein (that removeselectrons / protons from NAD);
3. Fewer electrons to electron transport chain / electron transport chain slows /stops / stops / slower oxidative phosphorylation;
4. Fewer protons for proton gradient;
5. Not enough ATP produced / energy supplied to keep cells alive / anaerobicrespiration not enough to keep cell alive;

Accept neurones require ATP for active transport of ions
Ignore references to resting potential
(c) 1. Mitochondrion has two membranes / inner and outer membranes;Accept cristae for inner membrane
2. For each (different) membrane a (different) carrier required;

Ignore reference to channel proteins
[7] (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
(b) 1. Triplet / three bases on mRNA;

1. Accept nucleotide for base
2. Accept DNA for mRNA
3. Ignore references to RNA unqualified
4. That code for an amino acid;
5. Accept code for stop / start
(c) (i) To join nucleotides together to form mRNA / premRNA / RNA;

Reject forming base pairs
Accept checking and correcting mismatched base pairs
(ii) Reverse transcriptase;

If they give two enzymes, no mark
(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
[6] (a) (i) UGC;
15
(i) TGCTAC;
(b) (DNA) contains introns / non-coding bases / mRNA only contains exons / codingbases;

Assume that 'it' refers to DNA
Neutral: DNA contains introns and exons
Neutral: 'splicing'
Neutral: pre-mRNA contains introns
Ignore refs. to start and stop codons
(c) Different primary structure / amino acid sequence / amino acid coded for;

Reject: different amino acids produced / formed
Neutral: refs. to bonds
(d) 1. Acetylcholine not broken down / stays bound to receptor;
2. $\mathrm{Na}^{+}$ions (continue to) enter / (continued) depolarisation / $\mathrm{Na}^{+}$channels (kept) open / action potentials / impulses fired (continuously);
3. (Intercostal) muscles stay contracted / cannot relax;
[7] (a) (i) Repeating units / nucleotides / monomer / molecules;
(ii) 1. $\mathrm{C}=$ hydrogen bonds;
2. $\mathrm{D}=\underline{\text { deoxyribose; Ignore sugar }}$
3. $E=$ phosphate;

Ignore phosphorus, Ignore molecule
(iii)

| Name of base | Percentage |
| :---: | :---: |
| Thymine | 34 |
| Cytosine / Guanine | 16 |
| Adenine | 34 |
| Cytosine / Guanine | 16 |

Spelling must be correct to gain MP1
First mark = names correct
Second mark $=\%$ correct, with adenine as 34\%
(b) (i) 153 ;
(ii) Some regions of the gene are non-coding / introns / start / stop code / triplet / there are two DNA strands;

Allow addition mutation
Ignore unqualified reference to mutation
Accept reference to introns and exons if given together
Ignore 'junk' DNA / multiple repeats
[8] (a) (i) Phosphate and ribose;

Accept in either order. Both correct for one mark.
For phosphate accept $\mathrm{PO}_{4} \mathrm{Pi} /(\mathrm{P})$ but not $P$.

Do not accept phosphorus.
Ignore references to pentose / sugar.
(ii) TAGGCA;
(b) (i) Does not contain hydrogen bonds / base pairs / containscodons / does not contain anticodon / straight / not folded / no amino acid binding site / longer; Assume that "it" refers to mRNA.

Do not accept double stranded.
(ii) (pre-mRNA) contains introns / mRNA contains only exons;Assume that "it" refers to pre-mRNA.

Accept non-coding as equivalent to intron.
(c) (i)

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

One mark for both figures correct
(ii) 1. Have different (base) sequences / combinations of (bases);
2. (Pre-mRNA) transcribed from different DNA / codes for different proteins;
[7] (a) (i) 9 ;

Accept: nine
(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.
(c) Number of bases

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

Second column correct;
Columns three and four correct;
(a) 1. Hydrolysis breaks proteins / hydrolyses proteins / produces amino acids (from proteins);
2. Protein synthesis involves condensation;
(ii) 1. Tissues / cells are being broken down;
2. RNA is digested / hydrolysed / broken down;
3. By enzymes from lysosomes;
4. New proteins not made / no new RNA made;
(e) 1. (RNA) associated with making protein;
2. New / adult tissues are forming;
(f) 1. In the first 6 days no / little oxygen supplied / with breakdown of tracheae, no /little oxygen supplied;
2. (Without tracheae) respire anaerobically;
3. Anaerobic respiration involves reactions catalysed by enzyme B/conversion of pyruvate to lactate / involves lactate production;
4. Enzyme A / Krebs cycle is part of aerobic respiration; Or, with emphasis on aerobic respiration:

1. Tracheae supply oxygen / after 6 days oxygen supplied;
2. (With tracheae) tissues can respire aerobically.
(a) Banding pattern changes as cheetah gets older / difficult to judge as tail is short / fluffy;

## 20

(b) (i) Mean not (always) a whole number;

Standard deviation not (always) zero;
(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
(c) Band width not the same on both sides of tail;
(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;
(b) Ile Gly Val Ser;
(c) (i) Has no effect / same amino acid (sequence) / sameprimary structure; Q Reject same amino acid formed or produced.

Glycine named as same amino acid;

$$
1 \text { It still codes for glycine = two marks. }
$$

(ii) Leu replaces $\mathrm{Val} /$ 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.
(d) (i) Interphase / S / synthesis (phase);
(ii) DNA / gene replication / synthesis occurs / longest stage;Allow 'genetic information' $=$ DNA.

Allow 'copied' or 'formed' = replication / synthesis
(a)

22

| DNA | $\boldsymbol{v}$ | 2 |
| :---: | :---: | :---: |
| mRNA | $\boldsymbol{x}$ | 1 |
| tRNA | $\boldsymbol{v}$ | 1 |

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

DNA polymerase is incorrect Ignore references to RNA dependent or DNA dependent Allow phonetic spelling
(b) (i) (Receptor / transcription factor) binds to promoter which stimulates RNApolymerase / enzyme X;

Transcribes gene / increase transcription;
(ii) Other cells do not have the / oestrogen / ERa receptors;

But do not accept receptors in general.
(c) Similar shape to oestrogen;

Binds receptor / prevents oestrogen binding;
Receptor not activated / will not attach to promoter / no transcription;
Accept alternative
Complementary to oestrogen;
Binds to oestrogen;
Will not fit receptor;
2 max
[6] (a) Will replace themselves / keep dividing / replicate;

Undifferentiated / can differentiate / develop into other cells / totipotent / multipotent / pluripotent;

Accept tissues
(b) Reverse transcriptase;

Allow phonetic spelling
(c) (i) Alters base / nucleotide sequence / causes frame shift;

Different sequence of amino acids in polypeptide / protein / primary structure alters the tertiary structure;

Accept any reference, such as adding bases, to changing the base sequence of the gene. Reject deletion / substitution. Idea of sequence essential so not makes different amino acids.
Accept answers involving stop / start codons and effect on protein.
(ii) Affects tumour suppressor gene;

Inactivates (tumour suppressor) gene;
Rate of cell division increased / tumour cells continue to divide; lgnore answers relating to oncogenes. May gain third point.

2 max
(d) Yes

SCID patients unlikely to survive / quality of life poor unless treated; Cancer that develops is treatable / only affects $25 \%$ / five children;

No
Risk of developing cancer is high / $25 \%$;
Cancer may recur / may not be treated successfully in future / only short time scale so more may develop cancer;

No mark for yes or no. Marks are for supporting argument based on biological reasoning.
Accept any points

