## Mark schemes

(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;
lgnore 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.

2
(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.
(b) 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
(c) 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.
(d) 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.
[15] (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.
(e) 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.
(f) 1. Enough DNA using PCR;
2. Compare DNA sequence with 'normal' DNA.
[15] (a) (i) (In all organisms / DNA,) the same triplet codes for the same amino acid;
5
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
(ii) 64;
(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
3. Prevents / changes splicing;
4. (So) faulty mRNA formed;

Accept exons not joined together / introns not removed
5. Get different amino acid sequence;
(a) Locus;

Accept: loci
(b) Differences in DNA / differences in base sequence of DNA;

Accept: number of different alleles / size/variation in gene pool
Reject: genes
(c) 1. Jack Russell (genetic) diversity is (significantly) greatest;
2. Bull terrier (genetic) diversity is (significantly) smallest / is mostinbred;
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 tosignificance;

Reference to significance must be relevant to examples given
(d) 1. (Bull terrier) breeding has included a genetic bottleneck/ smallpopulation/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/lessselection (pressure);
4. Increased number of different alleles/larger gene pool/more varietyof alleles;

Reject if genes used instead of alleles
Reject: lower frequency of alleles
Ignore ref to mutations
(a) 250000 ;
(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) One / an amino acid (can be) coded for by more than one triplet;
8
Accept codon for triplet
Accept description of triplet - $\underline{\text { three }}$ 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
(a) (i) 4 ;

9
(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);
3. Alters 3D structure on its own is not enough for this markingpoint.
4. Substrate not complementary / cannot bind (to enzyme / active site) / noenzyme- substrate complexes form;
(b) 1. Lack of skin pigment / pale / light skin / albino;
5. Lack of coordination / muscles action affected;
(c) Founder effect / colonies split off / migration / interbreeding;

Allow description of interbreeding e.g. reproduction between individuals from different populations
[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'.
(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;
2
[7] (a) (i) Phosphate and ribose;

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

1
(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 | $\mathbf{U}$ |
| :--- | :---: |
| 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) More that one polypeptide / chain;
12
Ignore references to haem / other groups
(b) (i) 141 ;
(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;
(c) Different primary structure / amino acids / different number of polypeptide chains;

Question is about haemoglobin so do not credit differences in DNA
(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
] (a) Banding pattern changes as cheetah gets older / difficult to judge as tail is short / fluffy;
13
(a) Bais
(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
(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;
[8] (a) Introns;
(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 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)

15

| DNA | $\boldsymbol{r}$ | 2 |
| :---: | :---: | :---: |
| mRNA | $\boldsymbol{x}$ | 1 |
| tRNA | $\mathbf{r}$ | 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.

1
[5] (a) (i) Deoxyribose;
16 pentose / 5C sugar = neutral
1
(ii) Phosphate / Phosphoric acid;
phosphorus $/ P=$ neutral
(b) Hydrogen (bonds);
(c) $381 / 384 / 387$;
(d) (Gln) Met Met Arg Arg Arg Asn;
(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
[8] (a) Phosphate;
17
Deoxyribose;
Q Candidates must specify deoxyribose. This term is a specification requirement.
Ignore anything that is not incorrect.
(b) 4;
(c) (i) 14 ;
(ii) 36 ;
If (c)(i) incorrect accept [50 - (c)(i)]
(d) Different genes;

Different (DNA) base sequences;
2
[7] (a) a length of DNA;
18 that codes for a single protein / polypeptide;
2
(b) by heating;to break the H -bonds (between complementary bases);
(c) (i) to allow the DNA polymerase to attach / start addition ofnucleotides / mark start and end of sequence to be copied / prevents strands re-joining;
(ii) because the sequences at the ends of the target sequence are different / one is at the beginning and one at the end;
(d) 8 ;

$$
\text { accept } 7
$$

(a) (i) actin (Accept tropomyosin);

## 19

(ii) myosin head;
(b) (i) $\mathrm{Ca}^{2+}$ 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) $\times 100$; (actual numbers are $10 / 18 \times 100$ )
(ii) sample not representative / large enough / individual muscle fibresdifferent sizes / contain different number of myofibrils;
(d) all some stain $=1$ fast dark and slow lighter $=2$
(e) change in base sequence in DNA / addition / deletion / substitution of a base in DNAof 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;
(a) (i) join / attach nucleotides, to form a strand / along backbone / phosphodiester bonds;
(reject reference to H bonds, complementary base pairing)
(ii) ribosome / RER;
(b) (i) CGTTACCAA;
(ii) CGU UAC CAA;
(c) substitution;
(d) (i) alanine;
(ii) (mutation 1) no change(to sequence of amino acids); codon for alanine / degenerate codon / same amino acid coded for;
(mutation 2)
(change in sequence) valine replaced by alanine / codon for alanine; folding / shape / tertiary structure / position of bonds may change; (reject peptide bonds)
[10] (a) high energy radiation / ionising particles;
named particles / $\alpha, \beta, \gamma$;
colchicine; x rays /
cosmic rays; uv (light);
carcinogen / named carcinogen;
mustard gas / phenols / tar (qualified);
(b) (i) removal of one or more bases / nucleotide;
frameshift / (from point of mutation) base sequence change;
(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;
(iii) blue and lilac; white;

| colour of petal |
| :---: |
| (white) |
| blue |
| lilac; |
| white; |

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 / fomation of new stop codon; different tertiary structure / different 3D structure / different polypeptide / shortened polypeptide;
different shape of active site / no active site present;
[5] (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;
(a) mutation changes the amino acid sequence / primary structure of Factor VIII protein;

24 changes the tertiary structure / 3D shape;
(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;
(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;

