4.1 GENETIC INFORMATION, VARIATION AND RELATIONSHIP - DNA, GENES AND CHROMOSOMES – MARK SCHEMES

Q1.

(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]

Q2.

(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]

Q3.

(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]

Q4.

(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]

Q5.

(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]

Q6.

(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]

Q7.

(a) More that 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]

Q8.

(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 <u>nucleotides</u> 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]

Q9.

(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]

Q10.

(a) (i) Phosphate and ribose;

Accept in either order. Both correct for one mark.

For phosphate accept PO_4 / Pi / Ⓟ 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]

Q11.

(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]

Q12.

(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 <u>and</u> ionic / disulfide bonds;
7. Quaternary structure is two or more polypeptide chains.

5 max

(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

[15]

Q13.

(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]

Q14.

(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]

Q15.

(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]

Q16.

(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]

Q17.

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]

Q18.

(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]

Q19.

(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]

Q20.

(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]

Q21.

(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]

Q22.

(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;
<u>white</u> pigment does not bind;
lilac pigment not produced / white pigment remains unchanged / enzyme 1 does not function;

4 max

(iii) blue and lilac; white;

colour of petal
(white)
blue
lilac;
white;

2

[9]

Q23.

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

1

(ii) myosin head;

1

(b) (i) 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) × 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]

Q24.

(a) 1. **One of** RNA / ribonucleic acid(s) / nucleotide(s)/nucleic acid(s) / rRNA / ribosomal RNA / ribosomal ribonucleic acid
and
one of protein(s) / polypeptide(s) / amino acid(s) / peptide(s) / ribosomal protein;

Reject DNA, deoxyribonucleic acid, tRNA, transfer RNA, transfer ribonucleic acid, mRNA, messenger RNA, messenger ribonucleic acid.

Ignore enzyme(s), base(s).

1

(b) 1. mRNA binds to ribosome;
2. Idea of <u>two</u> codons / binding sites;
3. (Allows) tRNA with <u>anticodons</u> to bind / associate;
4. (Catalyses) formation of <u>peptide</u> bond between **amino acids** (held by tRNA molecules);

5. Moves along (mRNA to the next codon) / translocation described;
Assume 'it' refers to ribosome.

3 max

(c) TGCGTAATA;
Any errors = 0 marks

1

(d) 1. Introns (in pre-mRNA);
2. Removal of sections of (pre-mRNA) / splicing;
Introns removed' scores 2 marks.
Reference to 'introns present in mRNA' disqualifies mp1 but allow ECF for mp2.
Accept for 1 mark mRNA contains <u>only</u> exons.

2

[7]

Q25.

(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]

Q26.

(a) 1. Hydrogen bonds between the base pairs holds two strands together

2. Many hydrogen bonds provides strength
Reject strong hydrogen bonds

2

(b) (Because) ribosomes assemble polypeptides using mRNA code
OR
DNA has two strands each with a different (complementary) base sequence;

1

(c) Codon;

1

(d) 1. (Because) some amino acids have more than one codon / mRNA code;

2. Correct example from table.

2

(e) 1. Stop translation;

2. Result in detachment of polypeptide chain from ribosome.

2

(f)

CAC	ATG	ACC
Val	Tyr	Trp

Mark each row

2

[10]