8.2 The control of gene expression (A-Level Only) - Gene mutation – Mark schemes

Q1.

(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 for amino 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'

6

(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

E > ***D*** > ***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

3

(c) **180**;

1

(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 <u>percentage</u> decrease in number of cancer cells / greater <u>proportion</u> of cancer cells killed

4. Greater / faster increase in number of healthy cells / more healthy cells replaced / divide / healthy cells replaced / divide faster;

Accept: converse for cancer cells

*For **differences**, statements made must be comparative*

3 max

(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;

2 max

[15]

Q2.

(a) 1. Replacement of a base by a different base (in DNA);

1

(b) 1. (Depends on) size / mass (of protein);

2. (Depends on) charge (of protein);

Accept for 2 marks 'Smaller / more highly charged move further'

2 max

(c) 1. Each protein has a different tertiary structure;

2. (Each) antibody has a specific antigen / binding / variable region / site;

3. So, (each antibody) forms different antigen-antibody complex
OR
(each antibody) only binds to complementary (protein);

3

(d) 1. Less NL3;

2. More NR2A **and** NR2B;

2

(e) 1. Higher ratio NR2B to NR2A with mutation;

Accept 'more' as equivalent to 'ratio'

2. (Perhaps) better memory in mice with mutation;

2

[10]

Q3.

(a) 1. Cell wall not formed / production inhibited;

*1. **Q** Accept: weakened cell wall, but do not accept 'cell wall is broken down'*

2. Lower <u>water potential</u> in bacterium;

2. Accept: converse

2. Must be clear that the lower water potential is in the bacterium

3. Water enters and causes lysis / expansion / pressure;

2 max

(b) Human cells lack enzyme (**B**) / have a different enzyme / produce different fatty acids / use different substrates;

Neutral: 'human cells do not have cell walls' as out of context

1

(c) 1. Change in base sequence (of DNA / gene) leading to change in amino acid sequence / primary structure (of enzyme);

1. Accept: different amino acids coded for

1. Reject: different amino acids produced

2. Change in hydrogen / ionic / disulphide bonds leading to change in the tertiary structure / active site (of enzyme);

2. Neutral: alters 3D structure / 3D shape

3. Substrate not complementary / cannot bind (to enzyme / active site) / no enzyme-substrate complexes form;

3

[6]

Q4.

(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]

Q5.

(a) (i) Does not code for amino acid/tRNA/rRNA;

Accept 'does not code for production of protein/polypeptide'

Reject 'that produces/makes amino acid'

1

(ii) Deletion mutation;

Accept 'deletion'

Ignore references to splicing

1

(b) (The) polymerase chain reaction;

Accept PCR

1

(c) 1. Probes are single stranded / have a specific base sequence;
2. Complementary base sequence on (specific) spacer

OR

3. Complementary/specific to (particular) spacer;
4. (In white squares probe) binds (to single-stranded spacer) and glows/produces light/fluoresce;

2. Need idea of complementary to spacer

3. Accept converse for dark squares

3

(d) 1. To see if strain is resistant to any antibiotics;
2. So can prescribe effective/right antibiotic;

OR

3. To see whether (any) vaccine works against this strain/ see which vaccine to use/ to produce specific vaccine;
4. (So) can vaccinate potential contacts/to stop spread;

OR

5. Can test other people to see if they have the same strain/ to trace where people caught TB;
6. Allowing control of spread of disease/vaccinate/treat contacts (of people with same strain) before they get TB;

Do not allow mix and match of points from different alternative pairs

2 max

[8]

Q6.

(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]

Q7.

(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]

Q8.

(a) 1 (DNA altered by) mutation;
2 (mutation) changes base sequence;
3 of gene controlling cell growth / oncogene / that monitors cell division;

4 of tumour suppressor gene;
5 change protein structure / non-functional protein / protein not formed;
6 (tumour suppressor genes) produce proteins that inhibit cell division;
7 mitosis;
8 uncontrolled / rapid / abnormal (cell division);
9 malignant tumour;

max 6

(b) cancer cells die / break open;
releasing DNA;

2

(c) normal DNA and changed DNA have different sequences;
DNA only binds to complementary sequence;

2

(d) fewer abnormal / cancerous cells / smaller tumours;
less cell damage / less spread / fewer locations to treat;

2

(e) mRNA base sequence has changed;
gene / DNA structure is different / has mutated;
cancer gene active / tumour suppressor gene inactive;

3

[15]

Q9.

(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]

Q10.

(a) secreted by the liver / storage / release from gall bladder into the duodenum / small intestine;
bile passes unchanged from small intestine to colon;

2

(b) (i) chance alone has not caused the difference (between the two patients types);
high steroid high bacteria (significantly) higher percentage of cancer patients / low steroids low bacteria (significantly) higher percentage of control patients;

2

(ii) some patients with low levels of one / both factor(s) have cancer;

1

(c) change in code / base sequence / structure of gene;
addition / deletion / substitution;
mRNA / transcription changed;
gene product / protein structure / amino acid sequence changed / different protein;
loss of function;
uncontrolled cell division;

4 max

[9]

Q11.

Essay Using DNA in science and technology

DNA and classification

2.2 Structure of DNA

2.3 Differences in DNA lead to genetic diversity

2.9 Comparison of DNA base sequences

Genetic engineering and making useful substances

2.5 Plasmids

5.8 The use of recombinant DNA to produce transformed organisms that benefit humans

Other uses of DNA

2.5 Cell cycle and treatment of cancer

5.8 Gene therapy;

Medical diagnosis and the treatment of human disease;

The use of DNA probes to screen patients for clinically important genes.