



A-Level Biology

Gene Mutations

Mark Scheme

Time available: 56 minutes

Marks available: 41 marks

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Mark schemes

1.

(a) Box 2.

An inversion will result in a change in the number of DNA bases.

Reject if more than one box with tick. Ignore crossed-out ticks

1

- (b) 1. (Increased) methylation (of tumour suppressor genes);
Accept abnormal methylation or hypermethylation
Ignore decreased acetylation of histones
2. Mutation (in tumour suppressor genes);
3. Tumour suppressor genes are not transcribed/expressed
OR
Amino acid sequence/primary structure altered;
Accept mRNA for transcription/transcribed
Accept tertiary structure altered
Accept different amino acid
Ignore reference to protein not being formed
4. (Results in) rapid/uncontrollable cell division;
Accept cell division cannot be regulated
Ignore growth

3 max

- (c) 1. Correct answer of $1.9/1.93 \times 10^{25} = 2$ marks;;
Accept $2 \times 10^{25} = 2$ marks
Ignore any numbers after 1.93
2. Incorrect answer but shows $84 = 1$ mark
OR
 $28 \times 3 = 1$ mark
OR
Incorrect answer but shows 672 divided by 8 = 1 mark;

2

[6]

2.

- (a) 1. Change in (sequence of) amino acid(s)/primary structure;
Reject amino acids are formed.
Reject amino acids code.
2. Change in hydrogen/ionic/disulfide bonds;
3. Alters tertiary/³ structure;
Reject active site.
Ignore quaternary.
Ignore 3D.

3

- (b) 1. Produce healthy (red blood) cells
OR
 Produce (normal) polypeptide/haemoglobin;
Produce only healthy (red blood) cells is only equivalent to mark point 1.
Accept produce 'normal'/non-SCD cells.
Ignore type of stem cell e.g. pluripotent.
2. No sickle/faulty/SCD (red blood) cells (produced)
OR
 No defective polypeptide/haemoglobin;
3. Stem/marrow cells (continuously) divide/replicate
OR
 Less chance of rejection (from brother/sister);
Differentiate is not equivalent to divide/replicate.
Ignore type of stem cell e.g. pluripotent.

3

(c)

Max 2 marks for marking points 1, 2 and 3

(For gene therapy)

1. No destruction of bone marrow
OR
 No destruction of stem cells;
Accept no destruction of faulty bone marrow unless context indicates this is against gene therapy.
2. Donors are not required;
*Stating 'only own cells used' is **not** equivalent.*
3. Less/no chance of rejection (own stem cells);

(Against gene therapy)

4. Sickle/faulty (red blood) cells still produced
5. Immune response against genetically modified cells/virus
OR
 Long-term effect not known (as is new treatment)
OR
 Virus could cause side effects;
Accept 'virus could cause problems' or 'risk(s) with virus'.

3 max

[9]

3. (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]

4.

- (a) Substitution;
Accept inversion or translocation
Ignore 'point mutation'

1

(b)

Max 2 marks for mark points 2, 3 and 4

1. ($VO_{2\max}$ **and** CS activity) increased for both groups;
2. No statistical test, **so** do not know if **differences** are significant
OR
 No statistical test, **so differences** could be due to chance;
Ignore standard deviation
Accept correct named statistical test eg t-test
3. Only 8 weeks training
OR
 Training did not last long;
4. Might not be true for all types of training/exercise/females;

3 max

(c) In Group C:

1. Less mitochondrial replication/production;
1. and 2. Accept converse for Group T
2. Less transcription (of genes) for mitochondrial proteins/CS
OR
Less translation of (mRNA into) mitochondrial proteins;
Accept less CS/enzyme is produced

2

(d)

Max 2 marks for mark points 3, 4 and 5

Ignore any answers relating to sample size or duration of investigation

Ignore 'correlation does not mean causation' unless qualified

For (no mark)

1. (From **Figure 2** Group T have) increased CS activity for Krebs cycle;
2. (from **Figure 1** Group T have) increased $VO_{2\max}$ **so** more oxygen (available)
OR
(from **Figure 1** Group T have) increased $VO_{2\max}$ **so** more aerobic respiration
OR
(from **Figure 1** Group T have) increased $VO_{2\max}$ **so** delayed anaerobic respiration;
Accept 'less lactate' for delayed anaerobic respiration

Against (no mark)

3. No correlation between (percentage change in) $VO_{2\max}$ and CS activity
OR
No correlation on **Figure 3**;
4. It might not be thymine causing the increase
OR
There may be other differences in the control region (of Group T) that cause the increase;
Ignore 'could be due to lifestyle/diet changes'
5. $VO_{2\max}$ /CS activity not the only measures of ability to exercise for longer;
Accept ideas that they did not measure ability to exercise for longer

3 max

[9]

5.

- (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]