

## Mark schemes

**1**

- (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]

- (a) 1. Cut (DNA) at same (base) sequence / (recognition) sequence;

*Accept: cut DNA at same place*

2. (So) get (fragments with gene) **R** / required gene.

*Accept: 'allele' for 'gene' / same gene*

2

- (b) 1. Each has / they have a specific base sequence;  
2. That is complementary (to allele r or R). *Accept description of 'complementary'*

2

- (c) 1. Fragments L from parent rr, because all longer fragments / 195 base pair fragments;

*Ignore: references to fragments that move further / less, require identification of longer / shorter or 195 / 135*  
*Accept: (homozygous) recessive*

2. Fragments N from parent RR, because all shorter fragments / 135 base pair fragments;

*1 and 2 Accept: A3 for 195 and A4 for 135*

2. *Accept: (homozygous) dominant*

3. (M from) offspring heterozygous / Rr / have both 195 and 135 base pair fragments.

*Accept: have both bands / strips*

*Reject: primer longer / shorter*

3

- (d) 1. (Cells in mitosis) chromosomes visible;  
2. (So) can see which chromosome DNA probe attached to.

2

- (e) (i) 1. For comparison with resistant flies / other (two) experiments / groups;

*Ignore: compare results / data / no other factors*

2. To see death rate (in non-resistant) / to see effect of insecticide in non-resistant / normal flies. *Accept: 'pesticide' as 'insecticide'*

*Accept to see that insecticide worked / to see effect of enzyme*

2

- (ii) (PM must be involved because)

1. Few resistant flies die (without inhibitor);
2. More inhibited flies die than resistant flies;
3. (PM) inhibited flies die faster (than resistant flies);

(Other factors must be involved because)

4. Some resistant flies die;
5. But (with inhibitor) still have greater resistance / die slower than non-resistant flies.

Accept: (with inhibitor) die slower than non-resistant flies

4 max

[15] (a) Reverse transcriptase;

3

1

(b) 1. Probe (base sequence) complementary (to DNA of allele A / where A is (and) binds by forming base pairs / hydrogen bonds; *Accept gene A*

2. So (only) this DNA labelled / has green dye / gives out (green) light;  
*Accept glows for green light*

2

(c) (i) 1. More probe binding / more cDNA / mRNA / more allele / gene A means more light;

2. DNA (with **A**) doubles each (PCR) cycle;

3. So light (approximately) doubles / curve steepens more and more (each cycle) / curve goes up exponentially / increases even faster;

3

(ii) (**G** because)

1. (Heterozygous) only has half the amount of probe for **A** attaching / only half the amount of DNA / allele A (to bind to); *Accept only one A to bind to*

2. (So,) only produced (about) half the light / glow / intensity (of **H**) (per cycle of PCR);

*If reference to 'half' for point 1, allow 'less light' in 2.*

2

[8] (a) (i) 1. Negative correlation;

4

*Accept: description for 'negative correlation'*

*Neutral: 'correlation'*

*Reject: positive correlation*

2. Wide range;

3. Overlap;

4. (Graph suggests that) other factors may be involved (in age of onset);

*2 / 3 Accept the use of figures from the graph*

*2 / 3 Can refer to age of onset or number of CAG repeats*

*Ignore references to methodology*

3 max

- (ii) 1. Age of onset can be high / symptoms appear later in life;  
*Accept: 'gene' for 'allele'*
- 2. (So) individuals have already had children / allele has been passed on;

**OR**

- 3. Individuals have passed on the allele / already had children;
- 4. Before symptoms occur;

2 max

- (b) (i) 1. Person **K**;
- 2. (As has) high(est) band / band that travelled a short(est) distance / (er) so has large(st) fragment / number of CAG repeats; *Must correctly link distance moved and fragment size*

2

- (ii) Run fragments of known length / CAG repeats (at the same time);  
*Accept: references to a DNA ladder / DNA markers*  
*Do not accept DNA sequencing*

1

- (iii) Homozygous / (CAG) fragments are the same length / size / mass;  
*Accept: small fragment has run off gel / travelled further*

1

[9

- 1 (a) 1. Carriers are heterozygous / have one normal copy and one mutant copy of gene /

**5** have one recessive allele / don't have the condition;

- 2. Both have DNA that binds (about) half / 50% amount of probe (that non-carrier does);
- 3. Probe binds to dominant / healthy allele so only one copy of exon in their DNA / have one copy of gene without exon / base sequence for probe to bind to;  
3. *Accept normal and gene*  
3. *Accept have a deletion mutation*

3

- (b) 1. Introns not translated / not in mRNA / (exons) code for amino acids / introns do not code for amino acids;

1. *Accept not expressed*

- 1. *Accept polypeptide / protein for amino acids*

- 2. Mutations of these (exons) affect amino acid sequences (that produce) faulty protein / change tertiary structure of protein;

2. *Accept deletion leads to frameshift*

- 2. *In this context, accept affects protein made*

3. So important to know if parents' exons affected, rather than any other part of

DNA / introns;

*Accept converse arguments involving - eg introns do not code for amino acids / proteins*

*Reject references to making amino acids, once*

3

- (c) 1. Restriction mapping / described;

2. DNA / base sequencing (of fragments) / description / name of method;

2

- [8] (a) 1. Closer the (amino acid) sequence the closer the relationship;

6

2. (Protein structure) related to (DNA) base / triplet sequence;

*Amino acid sequence is related to (DNA) base / triplet sequence = two marks;*

2

- (b) 1. Reference to base triplets / triplet code / more bases than amino acids / longer base sequence than amino acid sequence;

*Different (base) triplets code for same amino acids = 2 marks;*

*Degeneracy of triplet code = 2 marks*

2. Introns / non-coding DNA / degeneracy of code / more than one code for each amino acid;

*Ignore reference to codon.*

2

[4]

## Essay Using DNA in science and technology

7

### 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.

- (a) (i) To cut the DNA;

**8**

*Reject breakdown, cutting out*

1

- (ii) To separate the (pieces of) DNA;

1

- (b) Complimentary base sequence / complementary DNA; binds to both (haplotypes);

Label would show up in both;

*Idea of complimentarity required*

2

- (c) (i) Y chromosome inherited / comes from male parents / only found in males;

1

- (ii) Mitochondria in egg / female gamete / no mitochondria come from sperm / malegamete;

1

- (d) (i) Allows comparison;

Different (sized) areas covered;

2

- (ii) Wolves do not eat all of prey animal / do not eat (large) bones / skin;

Inedible parts make up different proportions / wolf eats different proportions;

2

- (e) Limited by food / prey; as prey increases so do wolf numbers / positive correlation;

Large range so other factors involved;

2

[12]

- (a) Restriction (enzyme / endonuclease);

**9**

1

- (b) Move towards anode / move because charged;

Different rates of movement related to charge / size;

2

- (c) (i) Piece of DNA;

Single stranded;  
Complementary to / binds to known base sequence / gene;

max 2

- (ii) DNA invisible on gel / membrane;  
Allows detection;

2

[7] (a) Mother and father both heterozygotes / Tt / carriers;

**10**

Probability of thalassaemia 1/4 and female 1/2;  
Probability of both 1/8;

3

- (b) (i) Cut at same base sequence as same enzyme used;  
Fragments are same length / size / have same charge;

2

- (ii) Single base occurs many times;  
Sequence of 20 unlikely to occur elsewhere;  
*Allow one mark for establishing the principle where neither marking  
point clearly made.*

2

[7] (a) Endonuclease / restriction enzyme;

**11**

1

- (b) DNA made of base pairs;  
Each base pair is same length / occupies same distance  
along backbone;

2

- (c) (i) Second blank box from left labelled 6;

1

- (ii) Distance moved depends on length / number of base pairs /  
second longest fragment / second shortest distance identified;

1

- (d) 5;

1

[6]

- (a) (i) Different genes / characteristics / features;

**12**

Reference to mutations;  
Or  
Base sequence determines protein;  
Different species have different protein sequences;

max 2

- (ii) Primer has different DNA sequence;  
DNA specific / complementary base-pairing; 2
- (iii) Electrophoresis separates DNA;  
(So they can be) identified by position on gel;  
Smaller / shortest fragments travel furthest / quicker / or  
reverse argument; 3
- (b) (*conventional*) Many lengths / all DNA / (*new*) one length;  
Each rung is DNA of one / specific length; 2
- (c) 1 Heat DNA;  
2 Breaks hydrogen bonds / separates strands;  
3 Add primers;  
4 Add nucleotides;  
5 Cool;  
6 (to allow) binding of nucleotides / primers;  
7 DNA polymerase;  
8 Role of (DNA) polymerase;9 Repeat cycle many times;

max 6

[15] (a) 1 (DNA altered by) mutation;

13

- 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] (a) (i) contains genes / nucleotides / sections of DNA / artificial

14

DNA from two species / 2 types of organisms;

1

(ii) carries gene / DNA (into the other organism / gene carrier);

1

(iii) expose cells to the fungus; non-resistant ones die, resistant ones survive;

OR identify by adding marker gene / gene probe / (qualified) marker probe; description of positive result

e.g. radioactivity / fluorescence / complementary base pairing;

2

(b) EITHER 1 cut desired gene (from DNA) of oat plant;  
2 using restriction endonuclease / restriction enzyme;

OR 1 use mRNA from oat which will code for resistance;  
2 and use reverse transcriptase to form desired DNA;

OR 1 make artificial DNA with correct sequence of bases;  
2 using DNA polymerase;

3 cut plasmid open;

4 with (same) restriction endonuclease / restriction enzyme;

5 ref. sticky ends / unpaired bases attached;

6 use (DNA) ligase to join / ref. ligation;

7 return plasmid to (bacterial) cells;

8 use of Ca<sup>2+</sup> / calcium salts / electric shock; (if ref. to 'insulin' allow 5 max.)

max 6

[10] (a) probe will attach (to mutant allele);

15

attaches to one DNA strand; as a result of complementary base pairing; radioactivity detected on film / X-ray / by autoradiography (if mutant allele present);

4

(b) for gene is only active in mammary cells / only affects milk / easy to obtain product / product produced in large amounts / gene passed to offspring;

1

against long term effects not known / qualified reference to animal exploitation e.g. use of embryos / effect of inserted gene on other sheep tissues / genes;

1

[6] (a) Correct answer: 1.25;

*Ignore working*

**OR** (if wrong answer)

$$\frac{\text{measurement in } \mu\text{m}}{40000} / \frac{\text{measurement in mm}}{40} = 1 \text{ mark}$$

*125 but wrong order of magnitude = 1 mark*

2 (ii) **C** has myosin / thick (and actin / thin) filaments;

**OR**

**A** has only actin / thin (/ no myosin / no thick) filaments;

**1 max**

(b) When contracted:

Thick & thin filaments/myosin & actin overlap more;

Interaction between myosin heads & actin / cross-links form;

Movement of myosin head;

Thin filaments / actin moved along thick filaments / myosin;

Movement of thin filaments / actin pulls Z-lines closer together;

Displacement of tropomyosin to allow interaction;

Role of Ca<sup>2+</sup>;

Role of ATP;

*Allow ref. to 'sliding filament mechanism' /  
described if no other marks awarded*

**4 max**

(c) (i) 8 has DMD but 3 and 4 do not / 12 has DMD but 6 and 7 do not / neither parent has the condition but their child has;

*Allow parents 3 and 4 give 8, parents 6 and 7 give 12*

**1**

(ii) **4 AND 7;**

1 (iii) Parental genotypes: 6 = **X<sup>D</sup>Y** AND 7 = **X<sup>D</sup>X<sup>d</sup>**

**AND**

Gametes correct for candidate's P genotypes – e.g.

$X^D$  and  $Y + X^D$  and  $X^d$ ;

Offspring genotypes correctly derived from gametes e.g.

$X^D X^D + X^D X^d + X^D Y + X^d Y$ ;

Male offspring with MD correctly identified:  $X^d Y$ ;

Probability = 0.25 / correct for candidates offsprings genotypes;

*Accept 1/4 / 1 in 4 / 1:3 / 25%*

*NOT '3:1' / '1:4'*

4

(d) (i) No gene fragment **G**;

1

(ii) Only one copy of gene fragment **F**;

Male has only one X-chromosome / is XY

(c.f. female has two / is XX);

2

(iii) 10 has only one copy of gene fragment **G**;

10 has only one normal X-chromosome / has one abnormal /  
allele / has one X / is  $X^D X^d$  / is heterozygous;

11 has two normal X-chromosomes / has 2 normal alleles /  
is  $X^D X^D$  / has not got X / has 2 copies of (F and) G;

3

(e) (i) To prevent rejection / prevent antibody production vs. injected cells /  
injected cells have (foreign) antigen (on surface);

1

(ii) Shows effect of cells / not just effect of injection / not just effect of salt  
solution;

1

(iii) Only one person tested so far – need more to see if similar results  
/ need more to see if reliable;

Need to assess if new (dystrophin positive) muscle fibres are  
functional / if muscle becomes functional;

Can't tell how widespread effect is in the muscle / sample taken  
near injection site;

Need to test for harmful side effects;

Need to test if successful for other mutations of dystrophin gene;

Need to assess permanence / longevity of result/insufficient time allowed in investigation;

(In this patient) only small response / %;

Further sensible suggestion;

4 max

[25] (a) Cocaine (binding) changes shape of transporter/prevents dopamine binding;

17

*Reject references to active site*

Transporter cannot move (bound) dopamine (through membrane / protein / into cell);

Dopamine remains / builds up in synapses (leading to feelings of pleasure);

3

(b) (i) Polymerase chain reaction / PCR;

1

(ii) Single-stranded DNA;

*Reject reference to a single strand of DNA*

Bases / sequence complementary to DNA / gene to be identified;

(Radioactively / fluorescent) labelled so that it can be detected;

2 max

(c) Mutation changes base sequence of gene / DNA;

*Accept references to active site*

(Thus) changing amino acid sequence;

Changes tertiary structure / shape of protein/transporter;

Cocaine binding site changes/cocaine cannot bind;

Dopamine can still bind (and be transported);

3 max

[9]