## Mark schemes

(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'
(ii) Deletion mutation;

Accept 'deletion' Ignore references to splicing
(b) (The) polymerase chain reaction;

Accept PCR
(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) andglows/produces light/fluoresce;
5. Need idea of complementary to spacer
6. Accept converse for dark squares
(d) 1. To see if strain is resistant to any antibiotics;
7. So can prescribe effective/right antibiotic;

## OR

3. To see whether (any) vaccine works against this strain/ seewhich 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/ totrace 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
(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 foramino 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'
(c) 180;
(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 percentage decrease in number of cancer cells / greater proportion of cancer cells killed
4. Greater / faster increase in number of healthy cells / more healthy cellsreplaced / 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] (a) 250000 ;
3
(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) 1. Cell wall not formed / production inhibited;

1. Q Accept: weakened cell wall, but do not accept 'cell wall is broken down'
2. Lower water potential in bacterium;
3. Accept: converse
4. Must be clear that the lower water potential is in the bacterium
5. 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
(c) 1. Change in base sequence (of DNA / gene) leading to change in amino acidsequence / primary structure (of enzyme);

1. Accept: different amino acids coded for
2. Reject: different amino acids produced
3. Change in hydrogen / ionic / disulphide bonds leading to change in the tertiarystructure / active site (of enzyme);
4. Neutral: alters 3D structure / 3D shape
5. Substrate not complementary / cannot bind (to enzyme / active site) / noenzyme-substrate complexes form;
(ii) 1. Change in amino acid / (sequence of) amino acids / primary structure;
6. Reject = different amino acids are 'formed'
7. Change in hydrogen / ionic / disulphide bonds alters tertiary structure /active site (of enzyme);
8. Alters 3D structure on its own is not enough for this markingpoint.
9. Substrate not complementary / cannot bind (to enzyme / active site) / noenzyme- substrate complexes form;
(b) 1. Lack of skin pigment / pale / light skin / albino;
10. 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) 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;

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1 \text { 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);

## 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.
(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;
(c) normal DNA and changed DNA have different sequences;
DNA only binds to complementary sequence;
(d) fewer abnormal / cancerous cells / smaller tumours;less cell damage / less spread / fewer locations to treat;
(e) mRNA base sequence has changed;gene / DNA structure is different / has mutated; cancer gene active / tumour suppressor gene inactive;
[15] (a) secreted by the liver / storage / release from gall bladder into the duodenum / small
intestine;
bile passes unchanged from small intestine to colon;
(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;
(ii) some patients with low levels of one / both factor(s) have cancer;
(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;
[9] (a) (i) actin (Accept tropomyosin);
(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;
(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 fibres different 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;

