

DUNMAN HIGH SCHOOL Preliminary Examination Year 6

H2 Biology Preliminary Exam 2023 Mark Scheme

Paper 1: Multiple Choice Question

1	D	11	D	21	А
2	D	12	С	22	С
3	С	13	С	23	В
4	А	14	D	24	D
5	А	15	А	25	В
6	D	16	В	26	С
7	А	17	А	27	D
8	В	18	В	28	D
9	В	19	В	29	В
10	С	20	С	30	А

Paper 2: Structured Question

Question 1

- (a) Ref to Multiple cell types are found in the human capillary such as red blood [2] cells and endothelial cell [name at least 2 cell types];
 - which support the cell theory that living organisms are composed of cells;

OR

- Ref to the nucleus of white blood cell/endothelial cell contains DNA + presence of mitochondria for synthesis of ATP;
- which support the cell theory that cells are the basic unit of life;
- (b) [Describe] The radioactivity level increased from 60 to 80 arbitrary unit from 5 [4] to 10 minutes;
 - [Explain] Ref to **ribosomes were synthesising new proteins** using the **radioactive amino acids** which contributed to the initial high level of radioactivity;
 - [Describe] The radioactivity level decreased from 80 to 5 arbitrary unit from 10 to 40 minutes;
 - [Explain] Ref to the **newly synthesised proteins entered the ER lumen** and subsequently moved to Golgi apparatus;

- (c) Some radioactive proteins synthesised are not secretory proteins hence [2] synthesized by free ribosomes;
 - some radioactive proteins are rER enzymes and remained in rER;
 - Some of the newly synthesised radioactive proteins have not reached Golgi apparatus at 40 minutes;

Any 2

- (d) (i) Peptidoglycan <u>cell wall</u>;
 - **Circular** DNA;
 - <u>70S</u> ribosomes;

Any 2

- (ii) Primary lysosomes fuse with phagosomes /vesicles containing bacteria taken in via phagocytosis;
 - Autophagy in which primary lysosomes fuse with vesicles containing worn-out organelles;
 - Lysosomes contain hydrolytic enzymes to hydrolyse DNA/ protein/ carbohydrates in bacteria and worn-out organelles;

Question 2

- (a) Unique **number and sequence of amino acids** determines bonds and [3] interactions of R groups;
 - Segments of polypeptide chain coiled into alpha-helices and folded into beta-pleated sheets + stabilized by hydrogen bonds between C=O and N-H groups of polypeptide backbone / peptide bonds;
 - Further folding into globular structure, stabilised by bonds between R groups – hydrogen bonds, disulfide bridges, hydrophobic interactions and ionic bonds (any 3);
- (b) An enzyme has an active site with a 3D conformation that is [2] complementary to the protein / polypeptide / peptide bonds it binds and acts on;
 - The **spatial arrangement / charge** of the **binding and catalytic residues** restricts the type of substrates it can catalyse;
 - As only certain substrates have chemical groups orientated in a manner that would allow formation of temporary bonds with the contact residues to form ES complex;
 - (Ref. to ES complex) Substrates must also have chemical groups that are orientated near catalytic residues to facilitate the breaking and reforming of bonds / conversion of substrate to product;

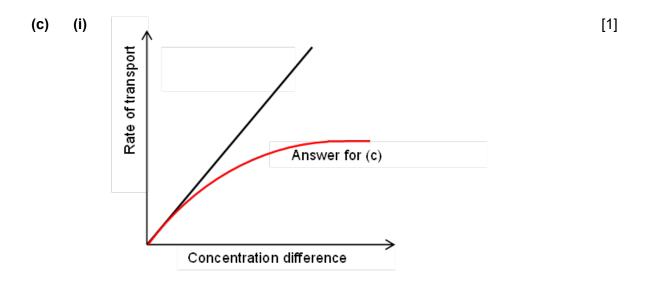
Any 2

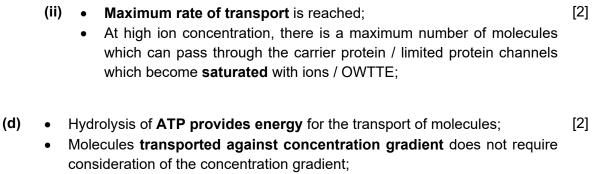
[2]

[3]

- (c) (i) Cooked protein is more digestible than raw protein + (Quote data) [1]
 Macia: 60% digestibility in raw while 70% in cooked OR NK8828: 56%
 digestibility in raw while 70% in cooked;
 - (ii) Cooked protein with acid is more digestible + (Quote data) Macia: [1]
 70% digestibility in cooked while 84% when cooked with acid OR
 NK8828: 70% digestibility in cooked while 80% when cooked with acid;
- (d) Cooking involves heating at high temperatures → High kinetic energy may [4] break the weak bonds eg. Hydrogen / hydrophobic interaction / ionic bonds (Any 1) present in kafirin;
 - Presence of high concentration of H+ in acid → neutralize negative charges and hence disrupt ionic bonds;
 - This results in protein **denaturation** / a change in 3D conformation;
 - Protease **can now easily access** and bind to the polypeptides → increased digestibility;

- (a) lons are **charged** and water molecules are **polar** / both molecules are [3] **hydrophilic**;
 - Unable to transverse the hydrophobic core of the phospholipid bilayer;
 - Hence, the **interior of channels are lined with hydrophilic residues** for the transport of water molecules and ions across;
- (b) Gated ion channel requires change in conformation to open it for [1] facilitated diffusion while water channel is constantly open and doesn't require conformation change;





• Bulk transport of molecules into or out of the cell does not take into account the concentration gradient;

Any 2

- (a) (i) Ref to most species of organisms use the same deoxyribonucleotides; [2]
 R: ref to genetic code being universal
 - 3D conformation of **active site** of *Taq* polymerase is **complementary** to the **3' OH** group of existing nucleotide;
 - (ii) Ref to specificity e.g. Allows for selective amplification of a segment of [2] DNA / flanking target sequence;
 - via complementary base pairing to <u>3'</u> ends of the template strands;

[2]

[2]

(b)

Feature	PCR	DNA replication		
Nature of primer	DNA primers provide free 3' OH for polymerase.	RNA primers used.		
How primer is synthesized	Primers synthesized artificially with knowledge of the target sequence.	Primers synthesized by		
Enzyme for polymerisation	<i>Taq</i> polymerase used for polymerisation / catalysis of phosphodiester bonds.	Human DNA polymerase used.		
Separation of template strand	Heat to 95°C to denature dsDNA.	Helicase used.		
Replication	Amplification of only the target DNA sequence.	Replication of entire DNA molecule.		
Location	Occurs artificially in a thermal cycler.	Occurs within nucleus of cells.		

Any 2

(c) (i) • Male 3;

- Only **Male 3 with genotype (29, 31)** would be able to contribute the chromosome with **29 repeats** at **locus E** to the child;
- (ii) Ref to only 5 gene loci were examined, (paternity testing requires at [1] least 13 different VNTRs given in preamble);
 - *Ref to* other males may have the same number of repeats at these 5 gene loci;

- (a) (i) Glucose can be used **directly** as a **respiratory substrate** to produce [3] **ATP**;
 - ...for binary fission to occur at a faster rate;
 - However, lactose needs to be first hydrolysed into glucose and galactose / monosaccharides, which takes time;
 - (ii) The allolactose in the cell binds to the active *lac* repressor, inducing a [3] conformational change and inactivate repressor;
 - Inactive repressor can no longer binds to the operator region of the *lac* operon;
 - hence RNA polymerase can access the promoter to initiate transcription of the *lacZ* gene to synthesise β-galactosidase;

[2]

- Only phage A can bind to the bacterial cells due to its tail fibre being complementary in shape to the receptors on the bacterial cell surface therefore can infect;
 - Hence can infect and cause osmotic lysis of the bacterial cell (which shows up as clear zones);

- (c) (i) In the experimental group, death rate was lower between 7-10% [accept: [3] average 8.25%] for all injection Day 0 to Day 3 than control group, with higher death rate between 50-55% [accept: average 52%] for all the injection days;
 - For injection Day 4, death rate in the experimental group increased sharply to 50%;
 - Bacteriophage treatment is effective in reducing death rate for only 3 days, after which another treatment is needed;
 OR

Bacteriophage treatment needs to be repeated every 3 days for death rate to remain low;

- (ii) Ref to Bacteriophage is more specific in targeting their harmful host [1] bacteria (complementary receptors), while antibiotics tend to have wider host range including beneficial bacteria.
 - Ref to Possible emergence of antibiotic resistant bacteria due to prolonged/inappropriate use of antibiotics, but no / low possibility of bacteriophage-resistant bacteria since they naturally infect bacteria.
 - Ref to Bacteriophages are harmless to chicken/human, while antibiotics in the chickens may trigger allergy response in human upon consumption.

Any 1

(a) 1. (In a diploid organism), both copies of the tumour suppressor gene must be [4] mutated so that no functional gene product can be produced, to result in abnormal cell proliferation OR
 If mutation occurs only in one copy of tumour suppressor gene, and the other

copy of tumour suppressor gene was able to code for **sufficient functional proteins** to prevent the cell from dividing abnormally;

- 2. Accumulation of mutations in several genes (involved in the control of the cell cycle) are required in a **single cell lineage** before the cell becomes cancerous;
- 3. Mutations may have occurred within **introns**, which are excised from the primary RNA transcript after translation, hence are non-coding;
- 4. The changed codon might still code for the **same amino acid** due to the **degeneracy of the genetic code**;
- 5. The changed codon might code for a different amino acid with a **similar R group** / **chemical property**, hence not affecting protein structure;
- The amino acid affected by the mutation may not serve a critical role in the protein which it is found in / non-essential amino acid, OR Mutations occurred in genes that do not regulate cell division;
- AVP, e.g., named specific checkpoints to arrest cell cycle when there are DNA mutations / DNA cannot be repaired;

(b) Any 2:

[3]

- 1. Blood stem cells can divide indefinitely via mitosis;
- 2. Blood stem cells are undifferentiated/unspecialised;
- 3. Blood stem cells are **multipotent**;

Compulsory:

4. So that they can differentiate into all blood cell types to replenish patient's blood cell supply;

- (c) (i) 1. As concentration of Paclitaxel increases from 5 to 50 nmol dm⁻³, the ratio [2] of cells in anaphase to those in metaphase decreases from 0.25 to 0.07;
 - 2. As concentration of Paclitaxel **increases** from **5 to 50 nmol dm**⁻³, the percentage of cells in mitosis **increases** from **5 to 38**;
 - (ii) 1. More cells are arrested in metaphase/cannot go past the M checkpoint; [2]
 - 2. As centromeres are not able to divide / spindle fibres cannot shorten, preventing movement of chromatids to opposite poles / AVP;

(a) (i) Epistasis;

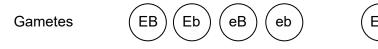
Marker's comments:

- There is no need to write dominant/recessive.
- Some still don't know what mode inheritance means? 'Autosomal inheritance' and 'dihybrid inheritance' are too vague.
- (ii) Eebb and EEbb;

[1]

[1]

(iii)Parental phenotypeBlackxBrown[4]Parental genotypeEeBbxEebb



Offspring genotype

	EB	Eb	eB	eb
Eb	EEBb	EEbb	EeBb	Eebb
	Black	Brown	Black	Brown
eb	EeBb	Eebb	eeBb	eebb
	Black	Brown	Yellow	Yellow

Offspring phenotype	Black	:	Brown	:	Yellow
Offspring phenotypic ratio	3	:	3	:	2

- (b) 1. Discontinuous variation;
 - 2. There are three distinct phenotypes (black, brown and yellow fur) and are nonoverlapping/do not form a continuum of intermediate shades unlike that of continuous variation;
 - 3. Only two genes contribute to melanin production unlike polygenic inheritance in continuous variation;

[3]

- (a) Differences:
 - 1. **Optimum temperature** of rubisco activase in cotton (30°C) is higher than that of false flax (25.5°C);
 - 2. **Maximum activity** of rubisco activase in false flax (0.118 arbitrary units) is higher than that of cotton (0.104 arbitrary units);
 - 3. **Range of temperature** that rubisco activase can function in cotton (from 20 to 42.5°C) is greater than that of false flax (from 20 to 37.5°C);

Similarities:

- Activity of rubisco activase in both plants increases, then decreases as temperature increases;
- 5. Activity of rubisco activase in both plants is **0.104 arbitrary units** at **30°C**;

Min. 1 similarity + 1 difference for max. 3m

- (b) 1. Less carbon dioxide is fixed with RuBP to form PGA;
 - 2. Less ATP is available to convert PGA to G3P and regeneration of RuBP from G3P (as it is used in the conversion of 2-phosphoglycolate to carbon dioxide);
 - During photorespiration, only 1 PGA is obtained per O₂ incorporated with RuBP, compared to 2 PGA when CO₂ is incorporated with RuBP. As PGA is required for regeneration of RuBP and for synthesis of sugar, rate of RuBP regeneration will be slower;

Any 2

(c) (i) (Glycolysis) + (Krebs' cycle) (net ATP + NADH) + (ATP + NADH + FADH₂) $(4 - 2 + 4 \times 2.5) + (2 + 6 \times 2.5 + 2 \times 1.5)$ (2 + 10) + (2 + 15 + 3)= 32 [2]

[2]

[3]

- (ii) 1. Some ATP could have been used to transport pyruvate / NADH / products [2] of glycolysis into matrix of mitochondrion;
 - 2. Some protons could have leaked across the intermembrane space without passing through ATP synthase;
 - 3. Some energy during the transfer of electrons along the electron transport chain could have been lost as heat;
 - 4. Glucose may not have been broken down / some intermediates are used in different metabolic processes;

[2]

Any 2

Question 9

- (a) P <u>glucagon;</u> Q <u>insulin;</u>
- (b) Both requires the activation of **relay proteins** in its signal transduction pathways; [3]

Both may result in the activation of **kinases** which leads to **cascade of phosphorylation**;

Both involves **2nd messengers** in its signal transduction pathways;

Both results in **signal amplification** where **more** relay proteins are activated compared to number of molecules of hormones bound to the receptors;

AVP;

Max 3

(c) Dimerization of the receptor OR change in conformation + exposes the tyrosine [2] kinase active sites on the intracellular domain of receptor;

Ref to **autophosphorylation** (A: cross-phosphorylation) of **tyrosine residues** on the intracellular tail;

(d) While they may activate the same cell signalling pathway, they may be **regulated** [1] **differently**, eg. there may be different rates of activation of IRS-1 relay protein / different rates of autophosphorylation of the receptor after ligand binding;

There may be **other** signalling pathways not shown which are not shared by the 2 ligands;

AVP;

Max 1

Question 10

(a) With the frequent / inappropriate use of antibiotics since its discovery, there has been [3] a rise in antibiotic resistance among bacteria;

Yet the number of new antibacterial agents discovered from 1983 to 2004 has been **decreasing from 16 in the period of 1983-87 to 3 in the period of 2003-04**;

[why looming?] Soon we will be left with no effective treatment against bacterial infections which can be deadly / AVP;

(b) Normal bacterial flora **beneficial** / does not cause disease while bacterial [1] pathogens cause disease by bringing harm to the host organism;

Insects have narrow temperature tolerance range, hence might die;

There may be migration of insects to higher altitudes;

There may be shift of insect activity range northwards and southwards;

Insects may have higher metabolic rates and hence higher activity rates;

Hence they may have shorter life-cycles;

They may be smaller in size if they grow faster;

There may be higher rate of mating and hence may increase reproduction rates, increasing their numbers and dispersion range;

[5]

AVP;

Any 5

Paper 3: Long Structured Question

Question 1

(a) There exists variation in attraction / aversion towards glucose among the [3] population of cockroaches;

The **use of insecticides select for cockroaches that are averse** / less attracted towards glucose;

These cockroaches **survive and reproduce**, **passing on the allele** for glucose aversion to their offspring;

Over time, the allele frequency for glucose aversion increases;

Max 3

- (b) (i) Maltose hydrolysed to 2α -glucose molecules + with the addition of a water [1] molecule, breaking of $\alpha(1 \rightarrow 4)$ glycosidic bond;
 - (ii) The <u>mean nuptial feeding duration of GA female is lower at 4s compared</u> [2] to WT females at 7s;

This is due to the release of glucose from the hydrolysis of maltose and other oligomers, which **deters the GA females** and results in the earlier termination of nuptial feeding / AVP;

(iii) The **GA** German cockroaches are expected to have **lower mating success**; [2]

And the **WT** German cockroaches are **killed by the glucose-containing baits of insecticides**;

(c) Lower salivary α -glucosidase activity at 70 au. compared to WT females' 130 au.; [2]

GA females hydrolyses nuptial secretion at lower rate, prolonging nuptial feeding duration, increasing reproductive success;

(d) (i) <u>inhibitor</u> of α -glucosidase;

[1]

(ii) No;

[3]

Acarbose is a <u>competitive</u> inhibitor which competes with oligosaccharides for the active site of α-glucosidase;

High dietary sugar consumption implies **high oligosaccharide concentration** in the intestines, which **overcomes the inhibitory effect of acarbose**;

(e) As the male German cockroaches increase in age, there could be increased [2] polyadenylation of the mRNA; The longer the 3' poly A tail, the longer the half life of the mRNA for translation:

The longer the 3' poly-A tail, the longer the half-life of the mRNA for translation;

OR

As the male German cockroaches increase in age, there could be increased synthesis of translation initiation factors; which stabilise ribosome assembly at 5' end of mRNA for higher rate of translation;

OR

As the male German cockroaches increase in age, there could be decreased ubiquitination of BGTG-1 protein; Hence protecting the protein from proteasome degradation;

OR

AVP mechanism; How it leads to more BGTG-1 protein;

(f) (i) Homologous;

[1]

- (ii) German cockroaches and *D. melanogaster* are likely to have **evolved from** [1] **a common ancestor**;
- (g) (i) The activation of mast cells leads to an inflammatory response / release of [1] histamine while activation of memory B cells leads to humoral response / release of antibodies;

AVP;

Any 1

(ii) IgE has variable region / antigen binding sites that are <u>complementary</u> to [2] allergen;

Constant region that is complementary to IgE receptors on mast cells;

Mast cells induced to release histamine;

Any 2

(h) <u>Class switching</u> where there is a change in constant region on the antibodies; [2]

The enzyme **activation-induced deaminase (AID) deletes** the C segments between 2 switch regions, resulting in a **different C segment expressed**;

Question 2

(a) Golgi apparatus; [1] (b) (i) Phenotype Antigens present on the red blood cells [2] A B H [2] Blood type A ✓ Image: Constraint of the red blood cells [2]

Blood type B		\checkmark	Correct with or without ✓
Blood type AB	\checkmark	\checkmark	
Blood type O			\checkmark

Any 2 rows correct for 1m

- (ii) 1 RBCs from AB donor / antigens A and B will be recognised as 'foreign' and [4] phagocytes such as macrophages / dendritic cells will phagocytose these cells;
 - 2 **B cell receptors** bind complementary to antigens A or B and engulf the donated RBCs via **receptor mediated endocytosis**;
 - 3 They will then hydrolyse / digest the cells, and present fragments / antigens on their surface MHC class II molecules;
 - 4 The **CD4**⁺ / **helper T cells** with the antigen receptors complementary to these antigens will be **activated**;
 - 5 The **B** / plasma cells will be activated to secrete antibodies against the A and B antigens;

Max 3 for points 1-5

- 6 The **antibodies** may bind to multiple AB donor's RBCs and result in agglutination / **aggregation of the RBCs**, which result in blood coagulation / clotting;
- (c) After transcription of the ABO gene sequence, the primary transcript undergoes [2] **splicing**; (Ignore: alternative splicing)

Introns were excised and exons 1 to 7 were spliced together;

(d) The <u>deletion</u> of a single nucleotide <u>G</u> in <u>exon 6</u> of the O allele;

Resulted in a frameshift mutation and the **early introduction of a stop codon** (nonsense mutation);

[4]

Leading to the synthesis of a truncated protein, that is **non-functional**;

Despite alleles A and B differing by 7 nucleotides and 4 amino acids, they both give rise to **functional proteins with different functions – adding different substrates to the H antigen** / AW;

Presence of both antigens A and B on the RBCs;

Max 4

- (e) (i) (As individual 8 is $I^{O}I^{O}$, the parents individuals 3 & 4 are $I^{B}I^{O}$. Hence the [1] genotypic ratio of their offspring will be 1 $I^{B}I^{B}$: 2 $I^{B}I^{O}$: 1 $I^{O}I^{O}$.) P($I^{B}I^{O}$) = 2/3 or 0.67
 - (ii) P(mother 7 being $I^{B}I^{O}$) = 2/3 or 0.67 [note: ECF awarded from e(i)] [1] P(father 6 being $I^{A}I^{O}$) = 1 P(son) = 0.5 P(blood type O from $I^{A}I^{O} \ge 10.25$

P(blood type O son) = 0.67 x 0.5 x 0.25 = 0.084 or 1/12

(a) Photosynthesis rate is measured as **amount of CO₂ taken in** for Calvin cycle; [2]

Net photosynthesis rate deducts the amount of CO2 released from respiration;

(b) (i) Heat stress treatment results in a reduction of mean net photosynthesis [2] rate from 12.8 to 8.2 μ mol CO₂ m⁻² s⁻¹;

The spraying of MeJA increases the mean net photosynthesis rate from 8.2 to 19.4 μ mol CO₂ m⁻² s⁻¹, reversing the effect of heat stress;

(ii) Spraying of MeJA increased chlorophyll production which leads to **increased** [3] **light absorption**;

This leads to higher rate of electron flow through the photosystems, resulting in **higher rate of ATP and NADPH synthesis**;

These lead to higher rate of Calvin cycle and hence higher rate of net CO₂ taken in for photosynthesis;

(c) (i)
$$t = \frac{|8.2-19.4|}{\sqrt{(\frac{0.79^2}{4} + \frac{1.4^2}{4})}} = 13.93 \text{ (2dp)}$$

working; final value to 2dp;

(ii) calculated t-value is greater than critical t-value;

implies that the net photosynthesis in wheat is **significantly different** after the spraying of MeJA among heat stressed crops;

[2]

[3]

Hence MeJA is effective in reducing heat stress among wheat;

Free Response Question

New 4a. There are many methods to classify organisms. Describe some methods and explain their limitations. [15]

Method 1: comparing morphological features

- 1. Several morphological features can be visually compared between organisms;
- 2. Organisms with similar structures are classified within the same group;
- For example, animals who warm-blooded and produce milk are classified as mammals. While animals who are cold-blooded, have dry skin covered with scales, and lay eggs are classified as reptiles / AVP any relevant example of grouping animals according to characteristics;
- 4. Organisms can be classified in a **hierarchical manner**, where they are first grouped based on a certain morphological trait, and **further grouped** based on another morphological trait;
- AVP: Ref to Kingdoms > Phylum > Class > Order > Family > Genus > Species (Any 3 in correct order);
- 6. Similar structures are likely to be inherited from a common ancestor;
- 7. AVP: On a phylogenetic tree / cladogram, those with similar structures will be **classified in the same clade**;

Limitations

- 8. It is not possible to classify organisms that had been **extinct with no complete fossil records**;
- 9. Some might have limited features for morphological comparisons Eg bacteria;
- 10. Comparison of morphological features is highly subjective and prone to errors;
- 11. Structures that look similar might not necessarily be inherited from a common ancestor or analogous structures might be mistaken as an homologous structure / Similar traits could be an indication of organisms adapting in a similar manner with respect to exposure to similar selection pressures;
- 12. Eg. Organisms with wings might be thought to have all inherited from a common ancestor, but birds and insects that fly had developed from different lineages / Placental and marsupial mammals looks very similar and might be mistaken as the same species, but they have in fact evolved from different ancestors / AVP any relevant example;
- 13. Structures might look too different to be considered homologous;
- 14. Eg. Whale's flippers and bat's wings;
- 15. AVP: Evolutionary relationship of organisms classified under the same clade may not be identified based on morphological traits. In other words, it is not possible to tell which organism within the same clade is more closely related to each other by merely comparing morphological traits;
- 16. AVP: Eg. Humans, Chimpanzees, Gorillas and Orangutans are all primates but it is difficult to classify further which organism is more related to each other, by merely comparison of morphological traits;

Method 2: comparing molecular features

- 17. DNA / RNA / polypeptide sequences of organisms can be aligned and compared;
- 18. Ref to chromosome painting;
- 19. Ref to DNA-DNA hybridization;
- 20. Ref to comparison of DNA banding patterns from gel electrophoresis / southern blot;
- 21. The amount of differences can be **quantified** / are **quantifiable**;
- 22. The more differences in sequence, the more distantly related the organisms are;
- 23. AVP: Hence classified under different clades evolved from different common ancestors in a phylogenetic tree;
- 24. Certain bases / amino acids might be conserved at certain positions;
- 25. These are likely to **indicate common ancestry** and have the organisms classified under the same group;

Limitations

- 26. It is difficult to classify organisms with no fossil records, or with fossil record poorly preserved such that **DNA cannot be extracted**;
- 27. AVP: Current technology only allows the comparison of short sequences, **whole genome comparison is still not available** / As a result, the classification might be based on comparison of **one or few genes** only, which might not be accurate;

Method 3: comparing biogeography

- 28. The **geographic distribution** of organisms over a period of time can suggest evolutionary relationship;
- 29. Based on the principle that each species originated only once,;
- 30. **Similar features** (morphological / molecular) observed between organisms can only be classified under the **same group** / **lineage** should they have **existed in the same geographical location** in the course of evolution;
- 31. Eg. Placental and marsupial mammals exist on different continents suggesting that they have evolved independently;

Limitations

32. Organisms which are similar might be found on different continents due to **continental drift** and be mistakenly classified into different groups / taxa;

QWC: 2 x [Identify method + describe (at least 1m) + explain limitation (at least 1m)];

4b. With reference to a named genetic disease, explain how genotypic changes can positively impact the survival of patients. [10]

1. Named genetic disease: **sickle-cell anaemia**;

Genotypic changes (mutations):

- 2. Single base substitution occurred in gene coding for β-globin chain in normal haemoglobin (HbA) (where thymine is replaced by adenine);
- 3. Codon on mRNA is changed;

Impact at molecular level:

- 4. Amino acid coded for changed from glutamic acid to valine;
- 5. Glutamic acid is **hydrophilic** while valine is **hydrophobic**;
- 6. Changing bonds formed between amino acid residues → change folding of protein
 → change 3D conformation / tertiary structure of haemoglobin;
- 7. At low oxygen concentration, the **hydrophobic portion** of abnormal haemoglobin (HbS) interact with each other, causing the molecules to **polymerise into fibres**;
- 8. Red blood cells changed from circular, biconcave shape to sickle-shape;

Positive impact on survival:

- Heterozygotes (Hb^AHb^S) have a higher reproductive success than homozygotes (Hb^AHb^A or Hb^SHb^S);
- 10. In malarial-stricken regions / in places where malaria exerts a selection pressure;
- 11. Hb^sHb^s homozygotes die of sickle-cell anaemia;
- 12. Because sickle-shaped red blood cells **obstruct blood vessels** and deprives multiple organs of oxygen, resulting in **organ damage**;
- 13. Sickle-shaped red blood cells are also more rigid and fragile, haemolysing readily;
- 14. **Hb^AHb^A homozygotes die** of malaria;
- 15. Because the malarial parasite (*Plasmodium*) can only infect normal red blood cells but not sickle-shaped ones;

QWC: 1 point from each heading;

5a. Explain how genetic variation can arise in nature. [15]

Independent assortment / random orientation

- 1. Independent assortment of homologous chromosomes during metaphase I;
- 2. Each homologue arranges itself on either side of the metaphase plate independently of the other pairs of homologous chromosomes;
- 3. Giving rise to different combinations of maternal and paternal chromosomes;
- 4. In **metaphase II**, the random orientation of **non-identical sister chromatids** along the metaphase plate is **independent** of other;
- 5. # Giving rise to different combinations of alleles in gametes;

Crossing over

- 6. Crossing over of homologous chromosomes occurs during prophase I;
- 7. # Giving rise to different combination of alleles on the chromosomes;

Random fusion

- 8. Random fusion of gametes during fertilisation;
- 9. Giving rise to genetically different zygote;

Mutations

- 10. 1 Gene mutations could occur due to insertion / deletion / substitution of base(s);
- 10. 2 Due to errors in DNA replication;
- 10. 3 Due to exposure to carcinogens;
- 11. Changing the sequence of nucleotides in a gene;
- 12. Giving rise to **new alleles** on chromosomes;
- 13. Chromosomal mutations could occur due to inversion / deletion / translocation / duplication;
- 14. Changing the structure of chromosomes / sequences of multiple genes / new alleles;
- 15. Chromosomal mutations could occur due to **non-disjunction**;
- 16. Changing the **number** of chromosomes;

Horizontal gene transfer (in prokaryotes) (max. 5m)

17. Horizontal gene transfer in bacteria could give rise to genetic variation;

- 18. In transformation, competent bacteria take up foreign DNA from the environment;
- 19. * Homologous recombination / insertion into host DNA genome can occur;
- 20. In **transduction**, **bacteriophages** transfer DNA fragments from **donor**/host **to recipient** bacterial cell;
- 21. Via generalised transduction due to error in packaging of bacterial DNA;
- 22. Via specialised transduction due to imprecise excision of prophage;
- 23. * Homologous recombination / insertion into host DNA genome can occur;
- 24. In **conjugation**, single-stranded copy of **F plasmid** is transferred from **donor (F⁺) to recipient** bacterial cell (F⁻);
- 25. Through conjugation tube;
- 26. Allowing **bacteria to gain new genes** which confer selective advantage (e.g., antibiotic resistance genes);

Antigenic drift and shift (in viruses)

- 27. Antigenic drift / shift can occur in viruses to give rise to new strains;
- 28. Lack of proofreading ability in RNA-dependent RNA polymerase in influenza or reverse transcriptase in HIV;
- 29. Results in high rate of mutation of viral genes;
- 30. **Two or more strains** infect the **same host cell** and the new strain contains a combination of gene segments occurs;

QWC: at least 2 valid points from any two of three sections: points **1 to 16**, points **17 to 26** and/or points **27 to 30**;

(#Mark points 5 and 7 only once) (*Mark points 19 and 23 only once)

5b. Compare the control of gene expression at the DNA and transcriptional level between prokaryotes and eukaryotes. [10]

DNA level

Similarities:

- In both organisms, the genes in tightly coiled / condensed regions are transcriptionally inactive, while genes in loosely coiled / condensed regions are transcriptionally active;
- 2. **Promoter regions** of genes to be silenced can be **methylated** in both organisms to **prevent binding of RNA polymerase**;
- 3. **Proteins associated** with DNA can be modified to **increase or decrease the level of DNA condensation** to decrease or increase level of gene expression, respectively;

Differences:

- 4. There exist **more intricate levels of control in eukaryotes** as the double stranded DNA is first coiled around histone octomers to form nucleosomes, where this bead-on-string structure is still transcriptionally-active with genes between nucleosomes accessible by RNA polymerase. Coiling of chromatin with 6-8 nucleosomes per turn results in the more tightly coiled 30-nm chromatin fibre that is **transcriptionally-inactive**;
- 5. However, in prokaryotes, DNA **simply supercoils** around non-histone proteins to be **transcriptionally inactive**.;

Transcriptional level

Similarities:

6. Gene expression can be positively regulated by **activator** proteins and negatively regulated by **repressor** proteins in both organisms;

Differences:

- 7. In prokaryotes, the regulatory proteins bind to sequences that are **close to or within the** promoter region;
- 8. While in eukaryotes, the regulatory proteins bind to **distal control elements** that are far away from the promoter of the genes to be controlled;
- 9. In prokaryotes, genes involved in the **same pathway** are controlled by a **single promoter** hence are simultaneously expressed with the activation of a single operon.;

- 10. However, in eukaryotes, genes involved in the **same pathway** have the **same control elements** and expressed simultaneously in the presence of transcription factors binding;
- 11. In prokaryotes, gene expression of each operon is controlled by **single type of activator and/or repressor proteins**;
- 12. While in eukaryotes, gene expression can be controlled by presence of **different types and combination of transcription factors**, enabling temporal and spatial regulation;

QWC: at least one comparison each at the DNA level and transcriptional level. At least one similarity and one difference.