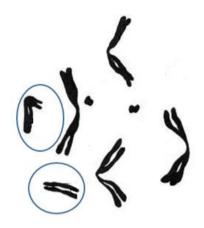


**1(a)** (i) Circle the sex chromosomes in Fig 1.1. [1]





(ii) Explain what is meant by 'heterogametic sex". [2]

- The organism where the pair of sex chromosomes are different in shape/size;
- Hence there are 2 types of gametes produced one type containing X chromosome, the other containing Y chromosome;
- (b) Explain how the behavior of chromosome during meiosis supports Mendel's law of segregation and independent assortment. [4] [Note: the basis is genes are found on chromosomes.]

Law of segregation:

- Separation of homologous chromosomes in <u>Anaphase I</u> /meiosis I and sister chromatids in Anaphase II/meiosis II;
- results in each gamete containing only 1 allele of each gene;

#### Law of independent assortment:

- Arrangement of pairs of homologous chromosomes across the equator in Metaphase I is random;
- Subsequent segregation results in random combination of maternal and paternal chromosomes in the gametes;
- (c) Describe one way in which translation of the maternal mRNA in the egg can be prevented. [2]

[Any 1 below]

- Translational repressors/ sequence-specific RNA binding proteins bind to the 5' UTR of mRNA;
- and prevents ribosome binding and formation of translational initiation complex; Or
- Binding of a **complementary RNA strand** to a specific region/critical region of the mRNA;
- to block the binding to ribosome;

(d) (i) Suggest an advantage of nuclei of the early embryo **sharing a common cytoplasm**. [1]

Any 1 below

- Large protein molecules present in the cytoplasm can move directly into the nuclei without having to cross any cell membranes;
- Sharing of a common pool of nutrients eg. nucleotides, ATP, enzymes, proteins;
- Enable nuclei to respond to the same signal;
- AVP;
- R: allows for nuclear divisions as the 3<sup>rd</sup> diagram (Fig 1.2) showed that there is no further division. There is only migration of nuclei to the periphery.
- (ii) Cellularisation occurs when cell membranes are formed around each nucleus to form cells.

Explain the importance of cellularisation in the continued development of the embryo. [2]

- Compartmentation which allows for setting up of specific environments;
- Allowing differentiation/ specialisation of cells via development of tissue specific structures/differential gene expression;
- Allows for more cells to be formed as cells undergo cell division/mitosis;
- Cells can then differentiate/specialised via expression of different genes/development of tissue specific structures;
- € Using the information from Fig. 1.3 and 1.4,
  - (i) account for the distribution of bicoid and nanos proteins. [2]
  - Bicoid and nanos <u>mRNA are only found concentrated</u> at the anterior and posterior <u>ends</u> of the embryo respectively;
  - When <u>translated</u>, the bicoid and nanos proteins will only be found in these regions respectively;
  - (ii) suggest explanations for the distribution of the hunchback and caudal proteins.[3]
  - [What is the distribution]Although mRNA concentration of both hunchback and caudal are high and distributed evenly (idea of) throughout the embryo, hunchback is present mainly in the anterior region while caudal is towards the posterior;
  - Bicoid protein can function as a repressor/ inhibitor for the translation of caudal mRNA;
  - Nanos can be a repressor for hunchback mRNA;
  - Very low concentration of nanos + caudal at the anterior end and bicoid + hunchback at the posterior end is due to diffusion;
  - AVP;
- (f) Suggest how the bicoid protein can result in the development of specific structures in the anterior region of the embryo. [3]
  - Bicoid protein act as a transcription factor/ activator;
  - Bicoid attach to promoter region of specific gene(s) and help recruit RNA polymerase to the promoter;

Results in transcription of specific genes that codes for specifc structures (eg. Mouth) found in the anterior region;
 A: If reference is made to bicoid being a repressor

A: If reference is made to bicoid being a histone acetylase

- (g) (i) If each of the DNA has undergone 10 rounds of DNA replication, how many DNA molecules are there in one polytene chromosome? [1]
  - [2<sup>10</sup> = 1024 x 2(because each polytene chromosome was formed from the replication of one homologous pair of chromosomes) =] 2048 DNA molecules;

(ii) Explain why it is unusual for homologous chromosomes to pair up in the salivary gland. [1]

 as salivary glands are somatic cells, not involved in the formation of gametes/ pairing of homologous chromosomes only occur in gamete forming cells / during meiosis;

(iii) Explain how repeated divisions can result in the banding pattern seen in Fig 1.5.[3]

- 1. idea that all the sister chromatids are formed as a result of semiconservative DNA replication and hence have **identical DNA sequences**;
- 2. Idea of DNA packaging depends on the DNA sequence hence same extent of packaging of DNA for same regions of all sister chromatids;
- Banding pattern Darker regions take up more stain as the DNA is more condensed around histones / more tightly packed / heterochromatin OR lighter region takes up less stain as the DNA is less condensed around the histones / loosely packed / euchromatin;

Scientists have labelled the banded regions of these giant chromosomes and use them to distinguish between different species of Drosophila.

- (h) (i) Suggest why different species of Drosophila show different banding patterns for the same polytene chromosome. [2]
  - Accumulation of independent mutations over time;
  - Different genes sequences are found / being expressed in different species (Reject different alleles);
  - (ii) Suggest a limitation of using polytene chromosomes for establishing phylogenetic relationship. [1]
  - Subjectivity in comparing extent of difference in banding pattern to determine relationship;
  - Small changes/ changes to a few nucleotides may not be reflected as a change in the banding pattern;

## **Question 2**

(a) Fig. 2.1 shows an event occurring in a dendritic cell infected with *Mycobacterium tuberculosis* (indicated with an \* in the figure). The arrowheads show this event involving several of the same organelle. B' is an enlargement of the boxed area.

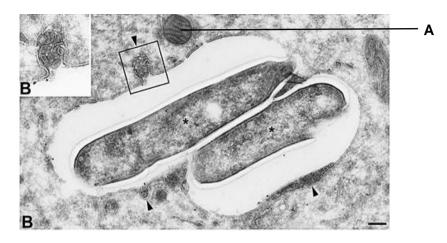


Fig. 2.1

- (i) Compare *Mycobacterium tuberculosis* with the structure labelled A, excluding size differences. [3]
- 1. Both contain ribosomes
- 2. Both contain circular DNA;
- 3. Both are membrane-bound/ consist of their own membrane;

		M. tuberculosis	A (mitochondrion)
4.	Number of membrane present/ presence of cristae	Single membrane/ lack     cristae	Double membrane / presence of inner membrane which is highly folded to form cristae
5.	Presence of (peptidoglycan) cell wall	• Yes	• No
6.	AVP	<ul> <li>rod-shaped</li> <li>bacterial/ prokaryotic cell</li> </ul>	<ul> <li>spherical shape here</li> <li>organelle found inside eukaryotic cell</li> </ul>

- (ii) With reference to Fig. 2.1, describe the event that is occurring in the dendritic cell infected with *Mycobacterium tuberculosis.* [1]
- 1. Fusion of lysosomes with the (membrane of) phagosome containing the bacterial cells;

- (b) Explain what happens to the IgG-coated beads when they are introduced to the macrophages. [3]
- 1. IgG are antibodies which opsonize the beads;
- 2. Macrophages have Fc receptors that bind to the antibody constant region;
- 3. Idea that this promotes / triggers phagocytosis or cause extension of long pseudopodia to engulf the IgG-coated beads;
- 4. Phagosome containing IgG-coated beads fuses with lysosome to form a phagolysosome/ to release hydrolytic enzymes;
- (c) With reference to Fig. 2.2,
  - (i) suggest how IgG-coated beads are being used as a control in this experiment. [2]
- 1. Phagosomes containing IgG-coated beads are successfully delivered to the lysosome causing a drastic drop in pH from 7.25 to 5.0 / rapid acidification to pH 5 and lower;
- 2. Serves as a positive control to show that contents in phagosome are normally acidified to provide specific pH for hydrolytic enzymes to work;
- 3. Reference to IgG-coated beads shows the normal response in change of pH after a foreign antigen/ pathogen is phagocytosed by macrophages;
  - (ii) explain the effect of *M. tuberculosis* in cells following phagocytosis. [3]
- 1. Following phagocytosis, phagosomes containing *M. tuberculosis* only acidify to pH 6.4/ slight drop in pH from 7.2 to 6.4;
- 2. Phagosomes containing *M. tuberculosis* fail to fuse successfully with lysosomes to lower the pH in its compartment;
- 3. Reference to *M. tuberculosis* not digested/ hydrolysed by hydrolytic enzymes;

[Total: 12]

# **Question 3**

- (a) Name one target cell of the cytokines. [1]
  - named innate immune cells (eg. neutrophils, dendritic cells, basophils, macrophages, mast cells etc)
- (b) With reference to Fig 3.1,
  - (i) explain how the structure of the receptor tyrosine kinase enable it to carry out its functions. [3]

Structure	Function	
complementary 3D shape of (extracellular) ligand binding site (Reject active site!)	enable specific binding of cytokine to initiate signalling pathway;	
contains (intracellular) tyrosine kinase domain JAK	can phosphorylate tyrosine residues on the receptor tail OR can phosphorylate relay protein STAT	
presence of tyrosine residues on receptor tails that can be phosphorylated (specific binding sites)	idea of phosphorylated tyrosine acting as 'docking'/binding sites for relay proteins (STAT);	
hydrophobic transmembrane region	Interact with hydrophobic core of membrane and enable RTK to stay embedded within membrane	

1m for correct structure  $\rightarrow$  function relationship. Max 3

- (ii) describe one instance in which signal is amplified in the cytokine signalling pathway. [1]
- Idea of many STAT proteins bind to phosphorylated tyrosines on the receptor tail to become activated /;
- (iii) describe two other ways in which the cytokine signalling pathway may be terminated, excluding the use of SOCS. [2]
- degradation of STAT dimer by degradative enzymes / proteasome;
- dephosphorylation / removal of phosphates from receptor tails / stat protein by phosphatases;
- disassociation of cytokine -> inactivation of tyrosine kinase domain (JAK)

any 2

(c) Recent findings on patients suffering from certain autoimmune diseases reveal mutations in the gene coding for SOCS.

An autoimmune disease is a condition where an individual's immune system attacks its own healthy tissue.

Explain how such mutations can lead to these conditons. [3]

- loss of function mutations in SOCS / SOCS becomes non-functional cannot inhibit JAK / JAK remains active / hyperactive JAK;
- STAT protein continues to be phosphorylated and activated -> formation of STAT dimer continues;

3. Leading to constitutive / continuous expression of target genes to produce proteins that stimulate excessive inflammatory responses (that may be harmful to own tissue);

# Question (Essay)

4 (a) Explain how evolution by natural selection can result in a decrease in the variation of a population and describe the various mechanisms that may increase or preserve this variation. [15]

Evolution by natural selection – decrease in variation

D: Definition of variation;

Variation (phenotypic) refers to the **differences in characteristics between individual organisms** belonging to the same natural population;

### (A) How natural selection decrease variation

Variation between individuals within a population

1. Idea of pre-existing variation in individuals within the same population;

Selective pressure

2. Environmental factors act as **selective agents** / **pressures**, selecting for individuals with the variations that allow them to adapt and outcompete others in the population.

#### Differential reproductive success

- 3. Individuals with certain variations that places them at an advantage will be selected against / will have selective disadvantage;
- 4. They will have lower chance to **survive and reproduce** / idea of lower reproductive success, giving rise to lesser offspring
- 5. Idea of less chance of passing down disadvantageous alleles to offspring;

Change in allele frequency (link to decrease in variation)

- The frequency of the alleles that are selectively favourable and passed down will increase in the next generation while those that are not favoured will decrease in frequency; OR proportion of individuals with alleles that confer selective advantage increases while the proportion of individuals with alleles that confer selective disadvantage decreases;
- 7. This leads to **changes in the allele frequencies in the gene pool** and over time, results in evolutionary changes and **microevolution**.

## (B) Mechanisms to increase variation

(Mutation and Gene flow)

- 1. Gene mutation results in **new alleles** and new characteristics. It **increases the gene pool** for natural selection to work on;
- 2. Chromosomal aberration results in **new genotypes with new combinations of alleles**, thus affecting the characteristics of individuals;
- 3. Idea that mutations must bring about changes which are **expressed in the phenotype** for other evolutionary processes to work on it.
- 4. Idea that only mutations that occur in **gametes** can be passed on to the next generation.
- 5. Gene flow is the movement of alleles from one population to another due to migration of individuals. If new alleles are introduced into a population, this can increase the genetic variation of the population.

### (C) Mechanisms to preserve variation

- 1. Heterozygote protection If the dominant phenotype is favourable, **recessive alleles in diploid organisms** can be preserved via the heterozygotes as they are masked by the dominant allele;
- 2. Balancing selection Balancing selection occurs when natural selection maintains two or more alleles in the gene pool of the population;
- 3. Eg. Of balancing selection: Heterozygote advantage If individuals who are heterozygous at a particular locus have greater fitness over both kinds of homozygotes, they exhibit **heterozygote advantage**, both alleles are maintained;
- 4. Eg of balancing selection: Frequency-dependent selection In frequencydependent selection, the fitness of a particular phenotype **declines if it becomes too common** in the population. Therefore, the **frequencies of the different phenotypes oscillate** over time;
- 5. Neutral variation Genetic variations that have **little or no impact on reproductive success** and thus natural selection does not affect these variations;
- Eg. Of neutral variation mutations that do not result in changes to proteins, and thus, no changes to phenotypes, do not have impacts on reproductive success; OR mutations that result in changes in proteins can also be neutral – eg. degeneracy of genetic code;

QWC: 1m for answering all 3 parts of the question.

**4 (b)** With reference to your knowledge of viral replication cycles, explain how phages can effectively eradicate bacterial infections in humans and explain the advantages and disadvantages of phage therapy over the use of antibiotics. [10]

## Phage replication cycle [max 5]

- R1 <u>Attachment</u>: The <u>tail fibres and base plate</u> of the phage recognise and bind to <u>specific</u> <u>receptors</u> on the cell surface of the host bacterium
- R2 The phage used in the phage therapy has to be specific to the bacteria that causes the infection in human;
- R3 Entry: <u>viral DNA enters host bacterium</u> through a hollow tube, leaving an empty capsid outside.
- R4 <u>Replication</u>: The phage <u>replicates its genome</u> and uses the bacterium's protein synthesis machinery to <u>synthesise phage enzymes and structural components;</u>
- R5 Thus effectiveness is in depleting/ competition for resources by making use of host energy, resources and cellular machinery, etc.;
- R6 Assembly: phage enzymes and other structural components <u>assemble</u> around the genome to <u>form mature phage particles;</u>
- R7 Exit: A phage-coded lysozyme breaks down the bacterial peptidoglycan cell wall to release the intact bacteriophages, causing cell <u>lysis</u> and <u>killing</u> the bacterium during exit.
- R8 <u>Each</u> of the newly produced phages can <u>infect a healthy bacterium</u>, and successive lytic cycles can <u>destroy an entire bacteria population</u> in just a few hours, thus effectively eradicate the bacteria that causes the human infection;

Antibiotic treatment (1mk)

 Antibiotics can kill the bacteria/ bacteriocidal or inhibits its growth/ bacteriostatic causes bacterial death by interfering with metabolic processes eg. cell wall synthesis / protein synthesis / cell membrane function / enzyme action;

Adv of phage therapy over use of antibiotics [max 4]

- A1 Target specific bacteria strains vs antibiotics which target most bacteria (non-specific)
- A2 Idea that beneficial bacteria in the body remains unharmed / only kill harmful bacteria in the body
- A3 Can kill antibiotic-resistant bacteria/ equally effective against antibiotic-sensitive and antibiotic-resistant bacteria
- A4 Can reduce the chances of generating antibiotic resistant bacteria through the over-use of antibiotics/ certain antibiotics are bacteriostatic, and as a consequence may more readily permit bacterial evolution towards resistance while phage therapy, bacteria are unable to regain their viability
- A5 Idea of auto-dosing Phages can increase in number over the course of treatment/ during the bacterial-killing process are capable of increasing in number specifically where hosts are located
- A6 Only one dose is required as phage can replicate in numbers to target more bacteria while antibiotic treatment has to sustain over period of time
- A7 Idea that single dose of phage therapy sufficient / low dosage used which may reduce cost (cheaper treatment)
- A8 Phages consist mostly of nucleic acids and proteins, they are inherently nontoxic

Disadv of phage therapy over use of antibiotics [max 2]

- D1 Phages can interact with immune systems, at least potentially resulting in harmful immune responses / adverse immune responses towards phages
- D2 Phage may mutate to become virulent / infectious towards human cells
- D3 Unknown health risks or allergies may arise due to introduction of phage into body
- D4 Not all phages make for good therapeutics, e.g. temperate phages
- D5 AVP?

Quality [1]: address both the replication cycle + at least 1 adv + 1 disadv of phage therapy

5	(a) Explain the advantages of having DNA as the hereditary material instead of RNA and describe the different roles played by RNA, including stable RNA-protein complexes in the cells. [15]
	<ul> <li>(A) <u>Advantages of having DNA as the hereditary material:</u></li> <li>[An important criterion of a good genetic material is to be able to preserve its genetic information and be able to pass this on to the next generation.]</li> </ul>
	1. <b>Double stranded</b> compared to RNA being single stranded;
	<ul> <li>2. Advantages of being <u>double stranded:</u></li> <li>a) Allows for accurate copying of genetic information as both strands act as a template for the synthesis of its complementary strand, resulting in daughter DNA molecules that are identical to the parental DNA;</li> </ul>

ł	<ul> <li>DNA repair – intact complementary strand can be used as a template to guide the repair of the strand with the error;</li> </ul>
	Since nitrogenous bases project into the helix, they are not exposed /are protected from mutagens, thus decreases the chance of mutation;
3. [	DNA is more <b>stable</b> than RNA
	<ul> <li>a) <u>Large number of hydrogen bonds</u> between nitrogenous base pairs across the two strands of polynucleotide that make up the DNA double helix;</li> <li>b) Hydrophobic interactions between the stacked bases</li> <li>c) Lower reactivity as there are no hydroxyl groups at carbon 2 of deoxyribose;</li> </ul>
	3. DNA molecule is larger – greater storage of genetic material;
4. /	<ul> <li>VP;</li> <li>DNA polymerase involved in DNA replication can proof read and hence they is less reading error compared to RNA polymerase;</li> </ul>
(B)	Roles of RNA
a)	<ul> <li>Messenger RNA (mRNA)</li> <li>mRNA carries the base sequence/genetic information of the polypeptide to be synthesised and act as a template for translation of the polypeptide;</li> </ul>
	• mRNA carries a copy of the genetic information from the nucleus to the ribosomes in the cytoplasm in the <u>eukaryotes;</u>
b)	Transfer RNA (tRNA)
	• tRNA can bind to an amino acid on its 3' end and has an anticodon that made up of that is complementary to a specific codon on the mRNA;
	• The main function of tRNA is to bring the appropriate/correct amino acid to the ribosome for protein synthesis/to pair to the codons on mRNA by complementary base pairing;
c <u>) F</u>	Ribosomal RNA (rRNA)
	• rRNA associates with ribosomal proteins to form ribosome;* (award once only)
	<ul> <li>rRNA of the small subunit can bind to the 5'UTR of the mRNA to allow for translation;</li> </ul>
	<ul> <li>ribozyme: peptidyl transferase – enzyme that catalyses the formation of peptide bonds between the amino acids of the polypeptide;* *(award once only)</li> </ul>
( <u>C)</u>	Roles of RNA-protein complexes 1. Ribosomes
	<ul> <li>Made up of rRNA and ribosomal proteins; *(award once only)</li> </ul>
	- Important in translation:
	<ul> <li>Provides the 3 sites Exit (E site), Peptidyl-tRNA site (P site) Aminoacyl-tRNA site (A site) for activated tRNA-aa complexes to bind in a complementary manner to the codons on the mRNA/ for the elongation of the polypeptide;</li> <li>Contains the enzyme peptidyl transferase – formation of peptide bonds between amino acids; *(award once only)</li> </ul>
	2. Spliceosomes

	<ul> <li>Made up of several snRNP – made up of proteins complexed with snRNA;</li> <li>Involved in excising introns and splicing exons to bind to form the mature mRNA</li> <li>Alternative splicing allows formation of different proteins via different combinations of exons from the same gene;</li> </ul>
	<ul> <li>3. Telomerase</li> <li>Made up of a <u>short RNA</u> template associated with an enzyme/ telomerase reverse transcriptase (TERT),</li> <li>Role: extend the <u>telomere length</u> during DNA replication/ synthesis of daughter strand, so as to postpone end replication problem;</li> </ul>
	<b>QWC</b> : [1] Answer both parts of the question AND at least 1RNA role and 1 RNA-protein complexes.
5	(b) Many processes in cells occur in a series of steps rather than a single step. With reference to different cellular processes, explain the advantages of such an arrangement in a eukaryotic cell. [10]
	<ul> <li>(1) <u>Process: Respiration</u> Note: Aerobic respiration consists of 4 stages – glycolysis, link reaction, Krebs cycle and oxidative phosphorylation. Each of these stage is made up of a multistep process.</li> </ul>
	<ul> <li><u>Controlled release of energy</u></li> <li>Gradual release of energy from oxidation of glucose (via glycolysis, link reaction, Kreb's cycle, oxidative phosphorylation) can be efficiently captured to form ATP/ controlled release of energy resulting in more efficient capturing of energy;</li> <li>If it is a 1 step process, amount of heat released may result in denaturation of enzymes/ controlled release so that heat so that <u>enzymes do not get denatured</u>; A: If reference is made to electron transport chain – electron carriers are arranged in progressively lower energy level to allow energy release to be used to pump H+ to from a proton gradient;</li> </ul>
	<ul> <li>Controlling rate of respiration/ ATP synthesis through controlling a specific enzyme</li> <li>Example: PFK (Phosphofructokinase)</li> <li>At high ATP concentration, PFK is inhibited by high ATP concentration, resulting in a decrease in the glycolysis;</li> <li>Prevents wastage cellular resources eg. glucose when energy is not required</li> </ul>
	<ul> <li>Intermediates/products can be channelled to alternative pathways</li> <li>When oxygen is not available, pyruvate (intermediate) can undergo anaerobic respiration/ lactate fermentation/ alcoholic fermentation so that glycolysis can continue to produce 2 net ATP to sustain cellular activity;</li> </ul>
	<ul> <li><u>Regeneration of acceptor for Krebs cycle to continue</u></li> <li>Krebs cycle consists of a number of steps such that its initial acceptor OAA is regenerated to allow the cycle to continue;</li> </ul>
	(2) <u>Process: Photosynthesis</u> Note: Photosynthesis consists of 2 stages – light dependent and light independent. Each of these stage is made up of a multistep process.

	<ul> <li><u>Non-cyclic and cyclic photophosphorylation</u></li> <li>Electrons from PSI can either reduce NADP or continue to the ETC from PSI</li> </ul>
	depending on the availability of NADP and/or the cells requirement for ATP;
	<ul> <li><u>Regeneration of acceptor for Calvin cycle to continue</u></li> <li>Calvin cycle consists of a number of steps such that its initial acceptor Ribulos bisphosphate is regenerated to allow the cycle to continue;</li> <li>[Award mark for regeneration once only – either in respiration or photosynthesis</li> </ul>
	Cell signalling Signal amplification
	<ul> <li>In multistep pathway, number of activated products is much greater than in the preceding step leading to amplification of signal;</li> </ul>
	A: example to illustrate the point – eg. production of cAMP/ phosphorylatic cascade
	Cellular response
	<ul> <li>Large cellular response can be obtained from a few extracellular sign molecules;</li> </ul>
	<ul> <li>Activation of specific relay proteins and/or receptors at each step lead to specif responses;</li> </ul>
	<ul> <li>Ref to ease in control and regulation of each step of the pathway;</li> <li>Allows for cross link between different signalling pathways – for a coordinate response;</li> </ul>
	<ul> <li>Termination of specific responses – idea of how specific enzymes could be inactivated in order to terminate certain responses only eg. phosphatase dephosphorylate specific kinases / phosphodiesterase convert cAMP to AMP;</li> </ul>
• • •	AVP
	Process identified must be a multistep process.