



# 2006 BIOLOGY Written examination 2

Solutions book

# This book presents:

- correct answers for Section A
- sample responses for Section B
- key knowledge information for Section B, relating each question to the coursework
- tips and mark allocations.

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# **SECTION A – Multiple-choice questions**

# Question 1

There are many levels of organisation in a chromosome. In eukaryotes, the DNA molecule coils around interacting proteins forming a supercoiled structure.

The name given to this supercoiled structure is

- A. chromatin.
- B. histone.
- C. nucleosome.
- D. centromere.

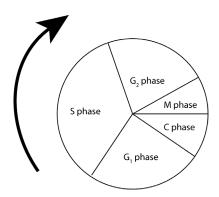
## Answer is A

Tips

- *A Chromatin is formed when interacting proteins package loops of coiled DNA into a supercoil.*
- **B** and **C** A nucleosome is formed when a section of DNA is wound around a core of eight histone proteins.
- **D** A centromere is the point of constriction on a chromosome which is required for chromosome movement during cell division.

# Question 2

The cell cycle describes the continuous sequence of events that takes place from one cell division to the next. The following diagram represents the life cycle of a cell.



The cell is most likely to be in interphase during phases

- A. M, C and  $G_1$ .
- **B. G**<sub>1</sub>, **S** and **G**<sub>2</sub>.
- C.  $G_1$  and  $G_2$  only.
- D. M and C only.

# Answer is B

- B In a cell cycle, interphase is the stage between nuclear divisions ( $G_1$ , S,  $G_2$ ).
- A, C and D M is associated with nuclear division and C with division of the cytoplasm.

The Cell Theory states that all cells are derived from pre-existing cells. Instructions for growth and development are passed from one generation of cells to the next through the processes of nuclear and cytoplasmic division. In many species, the instructions which determine their characteristics in successive generations are transferred in specialised cells known as gametes.

The process which gives rise to gametes is known as

- A. asexual reproduction.
- B. sexual reproduction.
- C. meiosis.
- D. mitosis.

# Answer is C

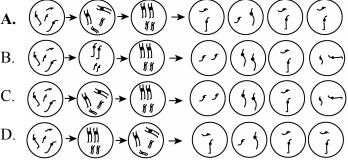
# Tips

- *C*-*Meiosis produces gametes cells that are haploid* (n).
- **D** is incorrect because mitosis produces diploid cells (2n), which are not gametes.
- *A* and *B* are incorrect because asexual and sexual reproduction are processes that give rise to new organisms, not gametes. Sexual reproduction can also refer to the fusion of gametes.

# Question 4

Meiosis is a cellular process which is also known as reductive division. During meiosis a very specific sequence of chromosome movements takes place to ensure that the resulting cells have all the genetic information they need to form a zygote.

Which of the following diagrams best summarises the correct sequence of events in meiosis?



# Answer is A

- A In the process of meiosis, a germline cell (2n) will produce four daughter cells (n). Each will have a single copy of one chromosome from each homologous pair. Before the cell divides into four daughter cells, the chromosomes in the cell undergoing meiosis will appear as double stranded, and synapsis will occur, resulting in a bivalent forming on the spindle.
- **B** and **C** do not demonstrate the correct configuration of chromosomes in the daughter cells (i.e. cells contain homologues and should not).
- **D**, while showing the correct configuration for daughter cells, does not demonstrate the correct order of chromosome movement in cell division. Double-stranded homologous chromosomes form a bivalent just prior to division, they do not dissociate.

All cells are programmed to divide, age and die, in a controlled manner within a given timeframe. Uncontrolled cell division leads to the accumulation of cancerous cells which form cancerous tissues. These tissues are either benign or malignant.

The process that normally results in the orderly, programmed death of cells is known as

- A. pinocytosis.
- B. apoptosis.
- C. necrosis.
- D. gametogenesis.

#### Answer is B

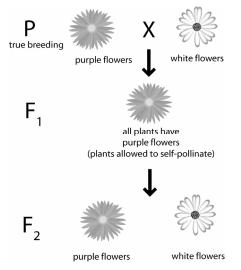
Tips

- B A poptosis is the process that results in the orderly, programmed death of cells.
- *A* is incorrect because pinocytosis is a form of bulk transport of liquid into a cell.
- *C* is incorrect because necrosis is the process of unprogrammed cell death.
- **D** is incorrect because gametogenesis is the process of generating gametes (meiosis).

#### Use the following information to answer Questions 6, 7 and 8.

While studying mathematics at the University of Vienna, the Augustinian priest Gregor Mendel carried out some breeding experiments on pea plants. The results of his study have formed the basis of the study of heredity.

In his experiments Mendel tracked heritable characteristics for three generations. The following represents one such experiment.



#### Question 6

In the pea plants used in the experiment, flower colour is a characteristic that has no variable form. Flowers are either purple or white.

True-breeding pea plants are also known as

- A. hybrids.
- B. monohybrids.
- C. heterozygous.
- D. pure breeding.

Answer is D

Tips

- **D** 'True breeding' is also known as 'pure breeding'. (When crossed among themselves, pure-breds always give rise to offspring identical to the parents for the gene under observation; they are homozygous for the gene.)
- *A* is incorrect because a hybrid is an organism produced by crossing two genetically diverse individuals, e.g. a mule (horse x donkey).
- **B** is incorrect because 'monohybrid' refers to a cross that involves one allele pair of characteristics.
- *C* is incorrect because 'heterozygous' refers to having two different alleles for a given genetic characteristic.

# Question 7

After allowing the  $F_1$  purple-flowered pea plants to self-pollinate, Mendel discovered that the  $F_2$  displayed a combination of purple-flowered pea plants and white-flowered pea plants.

These plants would have been produced in a ratio of

# A. **3 purple: 1 white**

- B. 3 white : 1 purple
- C. 1 white : 1 purple
- D. 1 purple : 1 white

# Answer is A

- A Correct ratio is 3 purple : 1 white. Two pure-breeding (homozygous) parents with different phenotypes will produce only heterozygous offspring ( $F_1$ ). If the offspring are permitted to interbreed to produce an  $F_2$ , the offspring will be produced in a phenotypic ratio of 3 (dominant) : 1 (recessive).
- **B** is incorrect because the phenotypes are reversed.
- *C* and *D* are incorrect because the ratios are incorrect.

# Question 8

If the genotype of pea plants that produce purple flowers is unknown, the most efficient way to determine the genotypes could be to cross a purple-flowered pea plant with

- A. an  $F_2$  generation purple-flowered pea plant.
- B. an  $F_1$  generation purple-flowered pea plant.
- C. a P generation purple-flowered pea plant.

# D. a P generation white-flowered pea plant.

# Answer is D

- D Crossing an F<sub>2</sub> purple-flowered pea plant with a pure breeding (homozygous)
  P generation white-flowered pea plant will be the best way to determine the genotypes.
  If there is a recessive allele in the genotype of the F<sub>2</sub> plants, it will be revealed by crossing with a homozygous recessive individual.
- *A*, *B* and *C* are incorrect because the proposed crosses with purple-flowered pea plants will not necessarily show if the purple-flowered pea plant is homozygous or heterozygous with respect to flower colour.

In humans there are four known blood groups. A person's blood group can be A, B, AB or O, where A and B indicate which antigens are found on the surface of the red blood cells. These four different blood groups result from various combinations of three different alleles of one gene.

The AB blood group occurs as a result of

- A. incomplete dominance.
- B. partial dominance.
- C. codominance.
- D. dominance.

#### Answer is C

Tips

- *C The AB blood group occurs as a result of codominance both alleles are fully expressed in the phenotype of the heterozygote.*
- *A* and *B* are incorrect because in incomplete dominance and partial dominance, heterozygotes display a phenotype that is a combination of both homozygous phenotypes.
- **D** is incorrect because in dominance, the dominant allele is expressed in the homozygous and the heterozygous form.

#### **Question 10**

In populations, a single characteristic can be controlled by more than one gene. In humans, skin pigmentation is controlled by at least three different genes and is an example of polygenic inheritance.

With respect to skin pigmentation, the human population demonstrates

- A. discontinuous variation.
- B. continuous variation.
- C. no variation.
- D. temporal variation.

#### Answer is B

- **B** The human population demonstrates continuous variation with respect to skin pigmentation.
- *A* is incorrect because discontinuous variation is characterised by a small number of distinct phenotypes due to the involvement of only one gene.
- *C* is incorrect because there is variation in skin pigmentation in the human population.
- **D** is incorrect because temporal variation refers to variation in season, time of day/night, which has no bearing on skin pigmentation in the human population.

The primary role of DNA in a cell is to determine what proteins the cell will make. RNA is a molecule that carries instructions from the DNA to the ribosomes where they are translated into proteins.

RNA differs from DNA in that it

- A. contains a deoxyribose sugar.
- B. is a shorter molecule than DNA.
- C. contains thymine as one of its nucleotide bases.
- D. is a double-stranded molecule.

# Answer is B

Tips

- **B** RNA differs from DNA in that it is a shorter molecule than DNA (due to the introns being removed).
- A is incorrect because RNA contains a ribose sugar.
- *C* is incorrect because uracil is a nucleotide base, not thymine.
- **D** is incorrect because RNA is a single-stranded molecule.

# Question 12

In the process of mRNA transcription, the part of the DNA to be copied is exposed. This strand is known as the template strand and the other strand of DNA

- A. is known as the sense strand.
- B. becomes the promoter.
- C. has the same sequence as the mRNA (with T bases instead of U bases).
- D. has the same sequence as the tRNA.

#### Answer is C

- *C The other strand of DNA has the same sequence as the mRNA (with T bases instead of U bases).*
- *A* is incorrect because the other DNA strand is the non-template or anti-sense strand.
- **B** incorrect because the promoter is the short strand of DNA (on the template strand) to which RNA polymerase binds to commence transcription.
- **D** is incorrect because the non-template strand will have T in its sequence, and tRNA will have U.

Gene mutations occur as a result of changes in gene sequences and are usually detected and repaired by enzymes. A point mutation is one kind of gene mutation and can

# A. involve the change of a single nucleotide base in a gene sequence.

- B. involve the addition of one or two nucleotide bases in a gene sequence.
- C. occur when a section of a chromosome breaks off and joins to another chromosome.
- D. occur as a result of non-disjunction in meiosis.

#### Answer is A

Tips

- A A point mutation involves the change of a single nucleotide base in a gene sequence.
- **B** is incorrect because point mutation involves the change (substitution/insertion (addition)/deletion) of only one base.
- *C* is incorrect because if a section of a chromosome breaks off and joins to another chromosome, more than a single nucleotide base will be changed. Therefore, by definition, it is not a point mutation.
- **D** is incorrect because non-disjunction leads to inclusion (or exclusion) of entire chromosomes in cells after cell division. It does not refer to mutation.

# Question 14

DNA replication is a very accurate process. Overall, mutations in individual genes are a rare occurrence. Certain environmental factors, known as mutagens, are capable of speeding up mutation rates.

Which of the following is **unlikely** to be a mutagen?

- A. mustard gas
- B. ultraviolet light
- C. nitrous acid
- D. polymerase

#### Answer is D

- **D** Polymerase is a group of enzymes responsible for joining monomers to form polymers; polymerases are not mutagens.
- *A*, *B* and *C* are incorrect because they are all known mutagens.

While all cells in an individual organism contain the same DNA, the cells of the organism can be quite different in their structure and function. Cells do not necessarily express all the genes of their genome simultaneously.

The expression of genes is **not** dependent on

- A. cell type.
- B. the stage of cell development.
- C. whether an organism is unicellular or multicellular.
- D. extracellular and intracellular conditions.

#### Answer is C

Tips

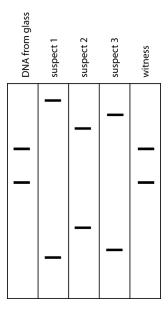
- C The expression of genes is not dependent on whether an organism is unicellular or multicellular.
- *A*, *B* and *D* are incorrect because expression of genes is dependent on all these options.

#### Use the following information to answer Questions 16, 17 and 18.

A daring daytime smash and grab robbery is carried out on a large jewellery store. The front window is broken and the contents of the window display stolen. Three individuals are seen running from the scene but disappear quickly down a nearby laneway. One of the thieves must have been cut by the broken glass as there is quite an amount of blood found on the broken shards. The blood is swabbed in order to obtain DNA. The samples are processed but the crime remains unsolved as the suspects have not been identified.

Four months later, following the offer of a large reward, a witness comes forward and identifies three suspects. DNA samples are taken from the suspects and the witness and compared with the DNA samples collected at the time of the crime. DNA can be used to identify an individual through a technique known as DNA profiling. The technique relies on the fact that a person's DNA is constant from cell to cell and, consequently, their profile is the same, regardless of what cells are used to obtain the DNA.

The results of the DNA profile are shown in the table below.



Constructing a DNA profile involves the use of two biotechnological techniques known as

- A. electrophoresis and transformation.
- **B. PCR and electrophoresis.**
- C. PCR and transformation.
- D. electrophoresis and DNA sequencing.

# Answer is B

Tips

- **B** Constructing a DNA profile involves the use of PCR and electrophoresis.
- *A*, *C* and *D* are incorrect because transformation and DNA sequencing are not techniques used in profile construction. Transformation is used in gene cloning. DNA sequencing is the process of determining the nucleotide sequence of a segment of DNA. Neither technique is used in DNA fingerprinting.

# Question 17

A DNA profile has been created for all of the individuals involved at the crime scene and the profiles will then be compared with the

- A. DNA of the store employees.
- B. DNA of the witness.
- C. DNA found on the glass.
- D. DNA of the suspects.

# Answer is C

Tips

- *C DNA* profiles will be compared with the DNA found on the glass as it is most likely that the DNA found on the broken window will be that of the perpetrator. A match between the DNA from the broken glass and the DNA profiles will identify the thief.
- Consequently, A, B and D are incorrect.

# Question 18

On the basis of the results of the DNA profile, the individual most likely to be convicted will be

- A. suspect 1.
- B. suspect 2.
- C. suspect 3.
- D. the witness.

# Answer is D

- **D** The individual most likely to be convicted will be the witness because both bands of their DNA match the DNA on the glass.
- *A*, *B* and *C* are incorrect because the bands for each suspect do not match the DNA on the glass.

Fossils are the preserved remains of organisms that lived on Earth in the geological past. The discovery of fossils is thought to have initiated the development of ideas about evolution. Fossils are commonly found in rock and their age can be determined using various techniques.

A reliable technique for dating fossils, known as absolute dating, assigns a numerical age to the fossil. Which of the following is **not** an example of an absolute dating technique?

#### A. use of index fossils

- B. radiometric dating
- C. electron spin resonance
- D. thermoluminescence

#### Answer is A

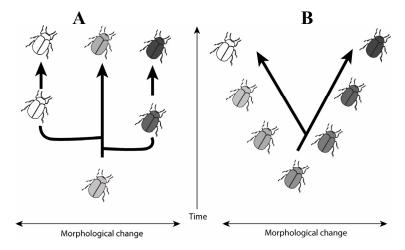
Tips

- *A* Use of index fossils is not an example of an absolute dating technique. It is an example of a comparative dating technique.
- *B*, *C* and *D* are incorrect as they are all examples of absolute dating techniques.

#### **Question 20**

Evolutionary trees commonly represent the descent of species from their ancestors as branches that sprout and diverge gradually, with each new species evolving continuously over long periods of time. However, palaeontologists rarely find gradual transitions of fossil forms of a species over time. In fact, species often appear suddenly as new forms. These new forms persist for some time and then disappear from the fossil record as suddenly as they appeared.

The diagram below is a representation of the two models of descent of species.



Which of the following is true of the diagram?

- A. Model A represents the model of evolution known as gradualism.
- B. Model A represents the model of evolution known as punctuated equilibrium.
- C. Model B represents the model of evolution known as punctuated equilibrium.
- D. Model A represents the model of evolution known as gradualism and model B represents the model of evolution known as punctuated equilibrium.

#### Answer is B

- B Model A represents the model of evolution known as punctuated evolution.
- A and **D** are incorrect as model A represents punctuated evolution, not gradualism.

• *C* and *D* are incorrect as model *B* represents gradualism.

Two populations of marsupial mice are separated from each other by an area heavily populated by humans. The two populations of mice are clearly different from each other and there are no individuals in either area which display characteristics of both populations.

It is reasonable to assume that

- A. the two populations are two separate species.
- B. the two populations could be the same species.
- C. the two populations are the same species.
- D. gene flow has occurred.

# Answer is B

# Tips

- B It is reasonable to assume that the two populations could be the same species. If they could produce viable fertile offspring when brought together, they are the same species.
- *A* and *C* are incorrect it is impossible to know for sure.
- **D** is incorrect because gene flow could not have occurred if the populations of mice are geographically separated from each other.

# Question 22

Chance events can cause allele frequencies in a population to change over time. When these random effects operate, the direction of the change is unpredictable and can differ from one generation to the next. The resultant pattern of change is known as genetic drift.

An example of genetic drift is

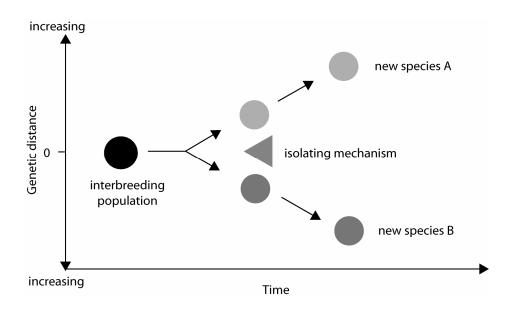
#### A. the loss of an allele from a population.

- B. the sum of all variation in a population.
- C. the phenotypic ratios in a population.
- D. the interbreeding of two populations.

# Answer is A

- *A An example of genetic drift is the loss of an allele from a population by chance. Genetic drift is a significant change in population gene frequencies which result independently of selection and migration, and are due to causes operating randomly with respect to the fitness of the alleles concerned.*
- *B*, *C* and *D* are incorrect because they do not meet these criteria.

Speciation is the process whereby a new species arises from a parent population. It occurs when the gene pool of the parent population is divided into two gene pools by an isolating mechanism. The two gene pools remain separate from each other and are no longer able to interbreed even when they are reunited.



Isolating mechanisms can operate before or after reproduction in individuals of a population. In animals, a post-reproductive isolating mechanism

- A. prevents mating and prevents the production of offspring.
- B. allows mating but prevents the production of offspring
- C. allows mating but prevents the production of fertile offspring.
- D. allows mating and allows the production of fertile offspring.

#### Answer is C

- *C In animals, a post-reproductive isolating mechanism allows mating but prevents the production of fertile offspring.*
- *A* is incorrect because post-reproductive isolating mechanisms do not prevent mating.
- **B** is incorrect because post-reproductive isolating mechanisms do not prevent the production of offspring.
- **D** is incorrect because post-reproductive isolating mechanisms do not allow the production of fertile offspring.

Primates are grouped in the order of mammals that share common characteristics and include monkeys and apes. Hominids are the group of primates that

- A. have tails.
- B. possess semi-opposable thumbs.
- C. show adaptations suited to arboreal life.

#### D. walk upright and have relatively large brains.

#### Answer is D

Tips

- **D** Hominids constitute the group of primates that walk upright and have relatively large brains.
- *A* is incorrect because hominids do not have tails.
- **B** is incorrect because hominids have fully opposable thumbs.
- *C* is incorrect because hominids are not arboreal.

#### **Question 25**

Evidence from cave paintings near Lascaux in France suggest that the Cromagnon people, an early form of *Homo sapiens*, worked co-operatively in their hunting practices to exploit the game herds they pursued. The development of more complex social behaviours and cultural evolution in early humans was attributed to larger brain size and the acquisition of language.

Development of cultural evolution did not include

- A. cultivation of crops.
- B. burial of the dead.
- C. hunting and gathering to obtain food.
- D. religious rituals.

#### Answer is C

- *C* Development of cultural evolution did not include hunting and gathering to obtain food.
- *A*, *B* and *C* are incorrect as they are all examples of cultural evolution.

# **SECTION B – Short-answer questions**

### **Question 1**

# AREA OF STUDY 1 – Heredity

#### Key knowledge – Patterns of inheritance in sexually reproducing organisms, transmission of heritable characteristics, variation

Students are expected to understand the patterns of inheritance in sexually reproducing organisms and be able to apply them to a specific example. Knowledge and understanding of inheritance associated with a single gene locus is essential. Ability to interpret the outcome of a monohybrid cross and distinguish between different types of gene expression is also required. Knowledge of phenotype and genotype and ability to make predictions about ratios of expected outcomes of crosses is essential.

In shorthorn cattle, coat colour is determined by one gene with two alleles.

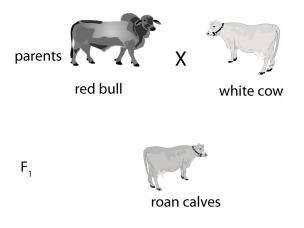
**1a.** What is an allele?

#### Sample response

An allele is an alternative form of a gene.

1 mark

The symbols for the alleles are red ( $C^R$ ) and white ( $C^W$ ). When pure breeding red cattle are crossed with pure breeding white cattle, the F<sub>1</sub> offspring have roan coats ( $C^R C^W$ ) which have a mixture of red and white hair. The genotype  $C^R C^W$  exhibits codominance in the phenotype.



**1b.** What is meant by codominance?

#### Sample response

Codominance is the relationship between two alleles of a gene such that both alleles are fully expressed in the phenotype of the heterozygote.

1 mark

The F<sub>1</sub> offspring are crossed.

**1c.** i. What are the expected phenotypes for coat colour in the  $F_2$ ?

#### Sample response

Three different phenotypes are expected in the F<sub>2</sub>: white, red and roan.

1 mark

**1c. ii.** What are the genotypes expected for coat colour in the  $F_2$ ?

#### Sample response

Expected genotypes in the  $F_2$  are  $C^W C^W$ ,  $C^R C^W$  and  $C^R C^R$ .

1 mark

1d. What is the expected ratio for genotype and phenotype in the  $F_2$ ?

#### Sample response

Expected ratio for genotype and phenotype in the  $F_2$  is 1 ( $C^W C^W$  – white) : 2 ( $C^R C^W$  – roan) : 1 ( $C^R C^R$  – red).

1 mark

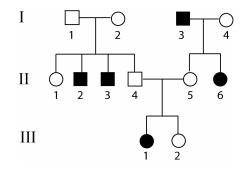
Total 5 marks

# **AREA OF STUDY 1 – Heredity**

#### Key knowledge – Patterns of inheritance in sexually reproducing organisms, transmission of heritable characteristics, variation

Students are expected to understand the patterns of inheritance in sexually reproducing organisms and be able to apply them to a pedigree. Knowledge and understanding of inheritance associated with a single gene locus is essential. Ability to assign alleles, interpret the outcome of a monohybrid cross and distinguish between different types of gene expression is also required. Knowledge of phenotype and genotype and ability to make predictions about probabilities of expected outcomes of crosses is essential.

In humans, earlobes can be attached or free. Earlobe attachment in a family was investigated over three generations and the data is recorded in the pedigree below. The shaded individuals have attached earlobes.



#### 2a. i. Is the attachment of earlobes a dominant or recessive trait?

#### Sample response

Attachment of earlobes is a recessive trait.

1 mark

**2a. ii.** Using the pedigree, explain the reason for your choice.

#### Sample response

Individual II-2 [or II-3 or III-1] has attached lobes even though their parents do not.

#### Marks

• 1 mark for naming individual II-2, II-3 or III-1

1 mark

**2a.iii.** Assign suitable allele symbols for this condition.

#### Sample response

Suggested alleles: A – free earlobe, a – attached earlobe

Tip

• The dominant trait must be assigned an upper case letter, and the recessive trait a lower case letter.

1 mark

18

**2b.** What is the genotype of individual II-1?

#### Sample response

Genotype of II-1: AA or Aa, it is not possible to distinguish either way.

1 mark

#### Tip

- Both possibilities must be included to earn the mark.
- **2c. i.** Use a Punnett square to demonstrate the outcome of a cross between individual II-4 and individual II-5.

#### Sample response

	(0.5) A	(0.5) a
(0.5) A	(0.25) AA	(0.25) Aa
(0.5) a	(0.25) Aa	(0.25) aa

#### Tip

• *The Punnett square must show the correct allelic composition of the gametes and proportions to gain the marks.* 

2 marks

2c. ii. What is the probability that their third child will be homozygous for free earlobes?

#### Sample response

The probability that their child will be homozygous for free earlobes is 0.25.

1 mark Total 7 marks

# **AREA OF STUDY 1 – Heredity**

#### Key knowledge – Patterns of inheritance in sexually reproducing organisms, transmission of heritable characteristics, variation

Students are expected to understand the patterns of inheritance in sexually reproducing organisms, in particular, sex-linked traits. Knowledge and understanding of inheritance associated with a single gene locus is essential. Knowledge of chromosome structure, cell reproduction and gamete production is essential.

Fragile X syndrome occurs in 1 in 1 500 male births and 1 in 2 500 female births. The mutation is often visibly expressed on the X chromosome and is due to an over-duplication of three nucleotides in the FMR-1 gene. In contrast with the normal 30 repeats of the CCG trinucleotide, individuals with fragile X syndrome can have 700 or more repeats within their allele.

**3a.** What is the name given to the position of a gene on a chromosome?

#### Sample response

The position of a gene on a chromosome is a locus.

**3b.** What is the term used to describe genes found on the X chromosome?

#### Sample response

Genes found on the X chromosome are X linked.

**3c.** Why is a male more likely to have fragile X syndrome than a female?

#### Sample response

A male is more likely to have fragile X syndrome because it is carried on the X chromosome. Males are hemizygous (they have only one copy of the X chromosome and will only have one allele for the trait), and if they inherit the recessive faulty gene on their X chromosome it will be expressed. Females are heterozygous for the trait and will only be affected if they inherit two recessive faulty genes.

#### Marks

- 1 mark for noting that males have only one X chromosome
- 1 mark for noting that females require two recessive genes to exhibit the trait

1 mark

1 mark

2 marks

Mutations occur in all organisms and can be somatic or germline in their nature.

**3d.** What is the difference between somatic and germline mutations?

#### Sample answer

A somatic mutation is a non-heritable mutation which occurs in a body cell of an organism and a germline mutation is a heritable mutation which occurs in cells that produce gametes by meiosis (germline cells). If/when a germline mutation is transmitted to a child, it is incorporated into every cell in the child's body whereas a somatic mutation is only acquired in a single body cell of one individual.

2 marks

#### Marks

• 1 mark for defining each kind of mutation correctly

Total 6 marks

# **AREA OF STUDY 1 – Heredity**

# Key knowledge – Molecular genetics, transmission of heritable characteristics, variation

Students are expected to understand the principal events involved in transcription and translation of DNA. Knowledge and understanding of the potential effects of a mutation on protein production is essential. Ability to read, understand and interpret a tabulated representation of the genetic code for DNA or mRNA.

Sickle-cell disease is an inherited blood condition. It is caused by the substitution of a single amino acid in the haemoglobin protein of red blood cells. Instead of normal beta haemoglobin, abnormal haemoglobin S (HbS) is produced. There are two alleles for the beta haemoglobin gene HbA (normal) and HbS (defective). When the oxygen content of an affected individual's blood is low, the HbS crystallises, deforming the red blood cells to a sickle shape. The inability to transport sufficient oxygen leads to tiredness, kidney or heart failure and children with the condition usually die by the age of 10 if it is left untreated.

4a. What is another type of point mutation that is not caused by a substitution?

#### Sample response

Another kind of point mutation not caused by a substitution is an insertion or a deletion, which leads to a frameshift mutation.

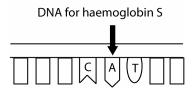
1 mark

	second base							_			
		U		С		A		G			-
	U	UUU UUC UUA UUG	phenylalanine phenylalanine leucine leucine	UCU UCC UCA UCG	serine serine serine serine	UAU UAC UAA UAG	tyrosine tyrosine stop stop	UGU UGC UGA UGG	cysteine cysteine stop trypotophan	UCAG	
first base	С	CUU CUC CUA CUG	leucine leucine leucine leucine	CCU CCC CCA CCG	proline proline proline proline	CAU CAC CAA CAG	histidine histidine glutamine glutamine	CGU CGC CGA CGG	arginine arginine arginine arginine	U C A G	third base
fire	Α	AUU AUC AUA AUG	isoleucine isoleucine isoleucine methionine/start	ACU ACC ACA ACG	threonine threonine threonine threonine	AAU AAC AAA AAG	asparagine asparagine lysine lysine	AGU AGC AGA AGG	serine serine arginine arginine	UCAG	
	G	GUU GUC GUA GUG	valine valine valine valine	GCU GCC GCA GCG	alanine alanine alanine alanine	GAU GAC GAA GAG	aspartic acid aspartic acid glutamic acid glutamic acid	GGU GGC GGA GGG	glycine glycine glycine glycine	U C A G	

Use the table of part of the genetic code (below) to answer parts **b** to **d**.

DNA for haemoglobin A





<b>4b.</b> What amino acid is normally produced in haemoglobin A?	
Sample response	
The amino acid normally produced in haemoglobin A is glutamic acid.	
	1 mark
4c. What is the mRNA triplet transcribed from the DNA from the HbS gene?	
Sample response	
The mRNA triplet transcribed from the HbS gene is GUA.	
	1 mark
<b>4d.</b> What amino acid is produced after the point mutation?	
Sample response	
The amino acid produced after the point mutation is valine.	
	1 mark

23

Total 4 marks

# **AREA OF STUDY 1 – Heredity**

# Key knowledge – Molecular genetics, tools and techniques, transmission of heritable characteristics, variation

In this question, students are required to have thorough knowledge and understanding of techniques used in gene therapy. Familiarity with the properties of tools and cells used in this process is essential.

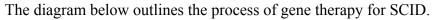
The condition severe combined immunodeficiency (SCID) is caused by a defective gene which disrupts the functioning of the B and T cells of the immune system. Children affected by SCID must live in completely sterile environments due to their susceptibility to disease and infection. The condition has been successfully treated by gene therapy.

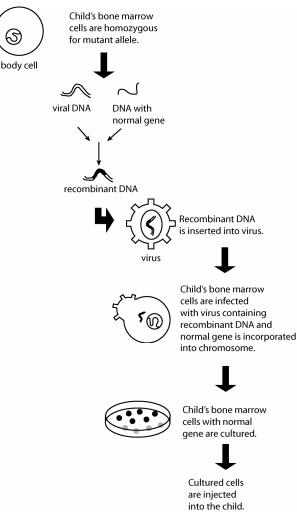
**5a.** What is gene therapy?

#### Sample response

Gene therapy is a technique which enables the delivery of normal functioning genes to individuals who have untreatable inherited conditions and at the same time has the potential to treat or cure the disorder.

1 mark





# **5b. i.** What is the role of the virus in this process?

#### Sample response

The virus acts as a vector.

1 mark

### **5b. ii.** Why is a virus well suited to this role?

#### Sample response

A virus is well suited to this role because it is capable of inserting a copy of its nucleic acid into the chromosomal DNA of its host. If the correct/functional form of a gene has been inserted in the viral DNA, it can be introduced and expressed by the injected cell as all cells produced from the injected cell will have the correct form of the gene in their DNA.

2 marks

#### Marks

- 1 mark for noting that the virus can insert a copy of its DNA into its host's cells
- 1 mark for noting that the virus will introduce the right DNA into the patient

In gene therapy for SCID, bone marrow cells are harvested, cultured and injected back into the child, who then produces functional B and T cells. The desired outcome is that the child demonstrates a normal immune response and remains healthy.

**5c.** Why are bone marrow cells ideal for use in gene therapy?

#### Sample response

Bone marrow cells are ideal for use in gene therapy because they are continually being produced. As a result, every mitotic division produces more cells with normal genes.

2 marks

#### Marks

- 1 mark for noting that bone marrow cells are constantly produced by the body
- 1 mark for noting that each mitotic division produces more cells with normal genes

**5d.** What is a limitation associated with the use of gene therapy?

#### Sample response

A limitation associated with the use of gene therapy is that it has not always been possible to ensure safe and effective delivery of therapeutic genes into cells and tissue or in the situation where a number of organs or body cells are affected, it may not be possible to insert functional genes into every organ or affected cell.

2 marks

#### Marks

- 1 mark for noting either that it may not be possible to introduce therapeutic genes safely and effectively
- 1 mark for noting that it may not be possible to insert the functional genes into all affected cells

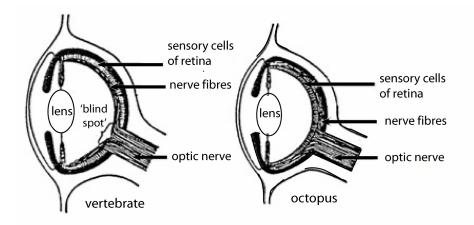
Total 7 marks

# AREA OF STUDY 2 – Change over time

#### Key Knowledge – Evidence of evolution, patterns of evolution

Students are expected, given an example, to demonstrate and justify their understanding of the difference between homologous and analogous structures and how they relate to convergent or divergent evolution.

The vertebrate eye and the octopus eye appear to be very similar in their structure and function. In the octopus eye, the sensory cells lie in front of the nerve fibres. The vertebrate eye however has an inverted retina which means that the sensory cells of the retina lie behind the nerve fibres. Consequently, the vertebrate eye has a blind spot where the optic nerve emerges from it.



#### **6a. i.** What is the term used to describe these similar structures?

#### Sample response

These are analogous structures.

1 mark

#### **6a. ii.** What is the meaning of this term?

#### Sample response

Analogous structures have similar function and appearance but demonstrate differences in their basic structure, which indicates that the two organisms have different ancestors. Vertebrates and octopus have different common ancestors, consequently, the structures presented here cannot be homologous structures.

2 marks

1 mark

#### Marks

- 1 mark for noting the similar function and appearance of analogous structures
- 1 mark for noting basic differences in their structures, meaning that the organisms have different ancestors

**6b.** What pattern of evolution is demonstrated by these structures?

#### Sample response

Convergent evolution

The platypus and the echidna are monotremes currently found only in Australia. Fossils of the ancestor of these animals have been found in Australia in Cretaceous rocks (138 million years BP – 65 million years BP). In Patagonia, South America, rocks formed from Palaeocene sediments (65 million years BP – 54 million years BP) revealed an upper right molar which bore close similarities to the tooth of the ancestral platypus, *Obdurodon*, which was present in Oligocene–Miocene deposits (40 million years BP – 23 million years BP) in Australia. The fine structure of the enamel and ridge patterns of the teeth in the Patagonian and Australian fossils were similar. South America and Australia were joined as part of one landmass which separated in the Cretaceous period (146 million years BP – 65 million years BP).

6c. What conclusions can be drawn about the fossils found in South America and in Australia?

#### Sample response

The Patagonian fossil suggests that the tooth-bearing platypus ancestor lived on Gondwana (South America/Australia) before the landmass separated in the Cretaceous. The South American line died out but the Australian line flourished and evolved to become today's population.

2 marks

#### Marks

- 1 mark for surmising that the platypus lived on Gondwana before the landmass separated
- 1 mark for concluding that the South American line died out and that the Australian line survived

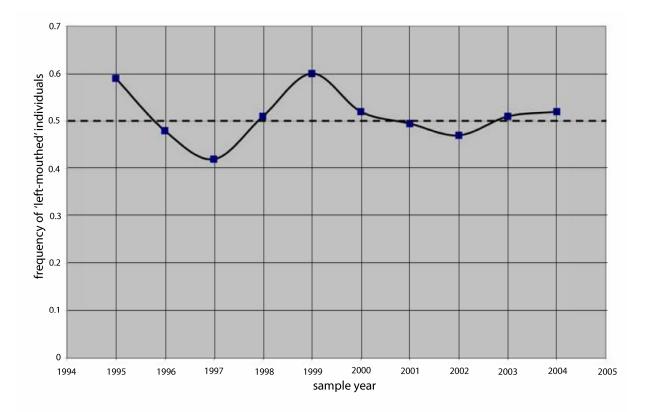
Total 6 marks

# AREA OF STUDY 2 – Change over time

# Key Knowledge – Change in populations, natural selection as a mechanism of evolution, evolutionary relationships.

In this question, students are expected to demonstrate a knowledge and understanding of polymorphisms as they relate to variation in populations. Students should be able to apply their understanding of natural selection and heterozygote advantage to explain the variations in relative frequencies of the cichlid population. In addition, an understanding of the significance of an ancestral trait and the ability to understand and interpret information to generate an evolutionary tree is essential.

*Perissodus microlepis* is a small cichlid fish that lives in Lake Tanganyika in Africa. This fish feeds on the scales of other fish by approaching their prey from behind to snatch scales from the flanks of their prey with their mouth. *P. microlepis* has an asymmetrical mouth which is oriented either towards the right or the left and they attack their prey from opposite sides, according to the position of their mouth. The 'right-mouthed' and 'left-mouthed' morphs occur in approximately equal numbers among individuals in a population. A 10-year study of 'left-mouthed' individuals recorded slight variations in relative frequencies. This trait is determined by simple Mendelian inheritance and is an example of a balanced polymorphism.



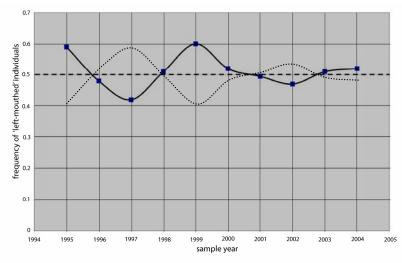
#### 7a. What is a polymorphism?

#### Sample response

A polymorphism is a variation in a trait which occurs as a result of more than one allelic form being present in the gene pool of a population.

1 mark

**7b.** On the graph above, plot the likely frequencies of 'right-mouthed' individuals over the 10-year interval.



#### Sample response



- Tips
  - The dashed line indicates the likely frequency of 'right-mouthed' individuals over the 10-year interval.
  - Note that the frequencies of both types of P. microlepis must total 1 at any point.

A hypothesis was proposed:

The fish, which *P. microlepis* preys upon, guard more effectively against attack from either the left side or the right side, depending on which *P. microlepis* morph is more common at the time.

**7c. i.** What mechanism is contributing to the balance of the cichlid population?

#### Sample response

The mechanism contributing to the balance of the cichlid population is natural selection or heterozygote advantage.

# Marks

• 1 mark for either response indicated

1 mark

7c. ii. Explain how this mechanism operates for *P. microlepis*.

#### Sample response

The majority of the prey does not guard against predators on the side that the less common morph attacks from. Therefore it has a feeding advantage that enhances its survival and reproductive success.

# Marks

- 1 mark for noting that the less common morph has a feeding advantage
- 1 mark for explaining why this is the case

2 marks Total 5 marks

# AREA OF STUDY 2 – Change over time

# Key Knowledge – Evidence of evolution, geological time, evidence of evolution, hominid evolution

Students are required to demonstrate their knowledge of appropriate dating techniques used for fossils of different ages and to provide evidence that they understand the reason for differences that may occur in fossils. An awareness of migration paths to Australia is also required.

An amniote is a vertebrate that develops a fluid-filled sac around an embryo. Amniotes are thought to have arisen from cotylosaurs. The fossil record provides no records of fluid-filled sacs surrounding developing foetuses. However, it is generally accepted that they evolved in the late Carboniferous period and were associated with the independence of amniotes from the water. The following table compares the estimated numbers of mutational changes (base substitutions) in the gene that encodes the respiratory protein, cytochrome c.

turtle	2.1 pigeon	2.4 duck	3.3 chicken	3.4 penguin	0 turtle
penguin	5.5	1.0	0.1	0	
chicken	5.4	0.9	0		
duck	4.5	0			
pigeon	0				

8a. Which species diverged most recently from the pigeon?

#### Sample response

The species that diverged most recently from the pigeon is the turtle – 2.1 mutational changes in the gene for cytochrome c.

1 mark

#### Marks

• 1 mark for 'turtle'

**8b.** What conclusion can be drawn about the presence of cytochrome *c* in all the amniotes listed?

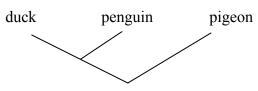
#### Sample response

The conclusion that can be drawn is that cytochrome c was present in the common ancestor and that is why it is present in all the amniotes.

1 mark

**8c.** Use the data from the table above to construct an evolutionary tree which demonstrates the evolutionary relationship between the pigeon, duck and penguin.

#### Sample response



#### Marks

- 1 mark for the structure of the tree
- 1 mark for the correct positioning of the birds

# Tip

• The evolutionary tree is based on numerical differences (mutational changes) between the pigeon, duck and penguin. The greater the numerical difference, the greater the evolutionary distance between the species.

2 marks Total 4 marks

# **Question 9**

# AREA OF STUDY 2 – Change over time

# Key Knowledge – Evidence of evolution, geological time, evidence of evolution, hominid evolution

Students are required to demonstrate their knowledge of appropriate dating techniques used for fossils of different ages and to provide evidence that they understand the reason for differences that may occur in fossils. An awareness of migration paths to Australia is also required.

There have been many human fossils found in Australia since 1884. Radiocarbon dating was used to date some of the skeletons and they were found to range in age from 9 000 years to 29 000 years.

Fossil location	Age (years)		
Lake Mungo (New South Wales)	26 000–29 000		
Keilor (Victoria)	13 000		
Kow Swamp (Victoria)	9 000–13 000		
Talgai (Queensland)	9 000–11 000		

Radiocarbon dating is suitable for fossil samples up to a particular age, beyond which it is difficult to accurately determine the level of carbon-14.

9a. i. What principle does radiometric dating depend upon?

#### Sample response

Radiometric dating depends on the fact that 'parent' elements contain radioactive isotopes which spontaneously decay or break down to form stable 'daughter' products.

1 mark

# 9a. ii. What alternative dating method would be appropriate for ageing these fossils?

#### Sample response

An alternative method for dating older fossils is potassium-argon dating.

1 mark

There were structural differences observed in the fossil skulls from these areas.

**9b.** Suggest two explanations for the differences in these skulls.

#### Sample response

#### **Explanation 1**

The differences between the skulls could be due to the normal expression of variation that exists within a population.

#### **Explanation 2**

The differences between the skulls could be due to the migration of two founder groups into Australia.

2 marks

Fossil evidence indicates that the earliest human inhabitants arrived in Australia about 60 000 years ago. These inhabitants are most likely to have migrated from South-East Asia.

9c. How did the first inhabitants get to Australia from South-East Asia?

#### Sample response

The first inhabitants arrived in Australia from South-East Asia across land bridges when the sea level was more than 150 metres lower than its current level. This occurred during an Ice Age – sea water was trapped in glaciers and ice masses.

1 mark Total 5 marks

# **END OF SOLUTIONS**