

2008

HIGHER SCHOOL CERTIFICATE

TRIAL EXAMINATION

BIOLOGY

General Instructions

- Reading time – 5 minutes
- Working time – 3 hours
- Write using black or blue pen
- Draw diagrams using pencil
- Board-approved calculators may be used
- *Write your Exam Number at the top of the pages*

Total marks - 100

Section I

75 marks

This section has two parts, Part A and Part B

Part A – 15 marks

- Attempt Questions 1- 15
- Allow about 30 minutes for this part

Part B – 60 marks

- Attempt Questions 16 - 29
- Allow about 1 hour and 45 minutes for this part

Section II

25 marks

- Attempt all parts of this question
- Allow about 45 minutes for this section

Section I**75 marks****Part A – 15 marks****Attempt Questions 1 – 15****Allow about 30 minutes for this part**

Use the multiple choice answer sheet.

Select the alternative A, B, C or D that best answers the question. Fill in the response circle completely.

Sample $2 + 4 =$ (A) 2 (B) 6 (C) 8 (D) 9A B C D

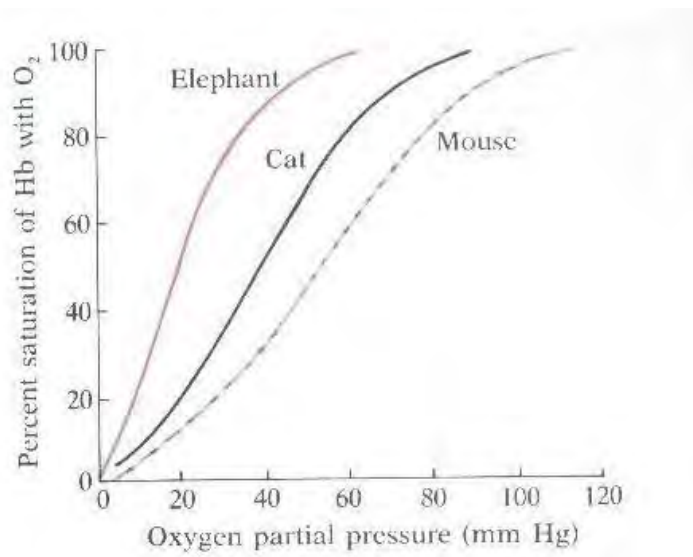
If you think you have made a mistake, put a cross through the incorrect answer and fill in the new answer.

A B C D If you change your mind and have crossed out what you consider to be the correct answer, then indicate this by writing the word *correct* and drawing an arrow as follows:*correct*A B C D

1. How is the majority of carbon dioxide transported in humans?

- (A) Dissolved in plasma.
- (B) Bound to haemoglobin.
- (C) In the form of hydrogen carbonate ions dissolved in plasma.
- (D) In the form of chylomicrons released into blood vessels.

2. The oxy-haemoglobin dissociation curves of three different sized animals are displayed in the graph.



What is the order of these animals in terms of the ease of haemoglobin losing oxygen in muscle tissue?

	haemoglobin offloads oxygen more readily	→	haemoglobin offloads oxygen less readily
(A)	elephant	cat	mouse
(B)	mouse	cat	elephant
(C)	cat	elephant	mouse
(D)	elephant	mouse	cat

3. Which regulatory processes explain the differences in the urine excreted by freshwater and marine fish?

	adaptations of freshwater fish	adaptations of marine fish
(A)	diffusion of salts from gills, diffusion of water into fish via gills, active transport of salt into fish via gills	diffusion of salts into gills, diffusion of water from fish via gills, active transport of salt out of fish via gills
(B)	diffusion of salts from gills, active transport of water into fish via gills, active transport of salt into fish via gills	diffusion of salts into gills, active transport of water from fish via gills, diffusion of salt out of fish via gills
(C)	diffusion of salts into gills, diffusion of water from fish via gills, active transport of salt out of fish via gills	diffusion of salts from gills, diffusion of water into fish via gills, active transport of salt into fish via gills
(D)	active transport of salts into gills, diffusion of water from fish via gills, diffusion of salt out of fish via gills	active transport of salts from gills, diffusion of water into fish via gills, diffusion of salt into fish via gills

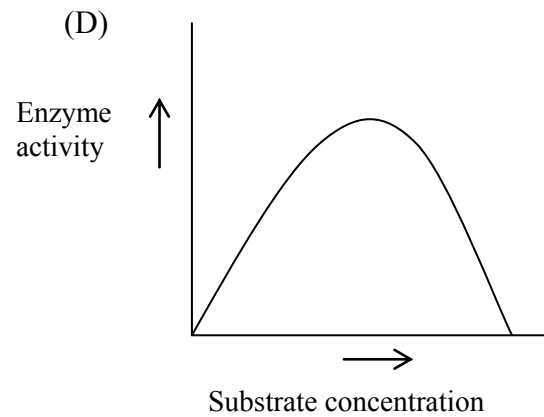
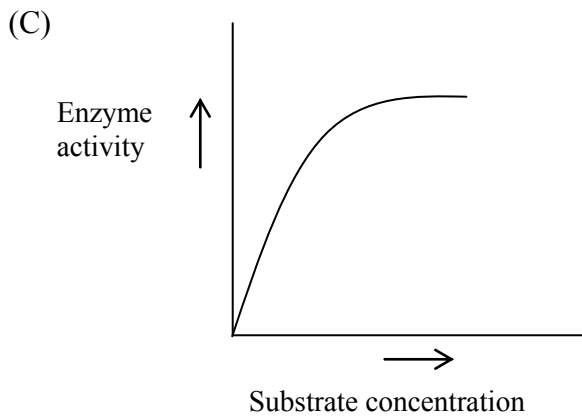
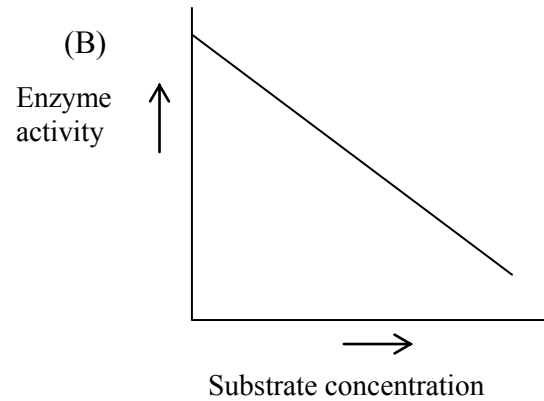
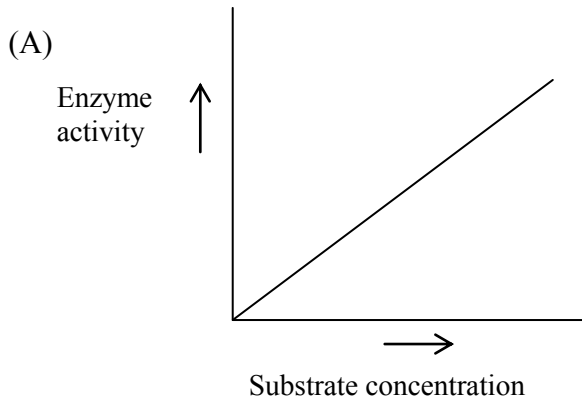
4. Red-green colour blindness is inherited as a sex-linked recessive condition. If a colour-blind woman marries a man with normal vision, what would be the expected phenotypes of their children?

- (A) All of their children will be colour-blind.
- (B) 100% of their daughters will be colour-blind.
- (C) 100% of their sons will be colour-blind.
- (D) About 50% of their sons and 50% of their daughters will be colour-blind.

5. Which scientist is credited with discovering sex-linkage by studying the inheritance of eye colour in fruit flies?

- (A) F. C. Crick.
- (B) T. H. Morgan.
- (C) R. C. Punnett.
- (D) W. S. Sutton.

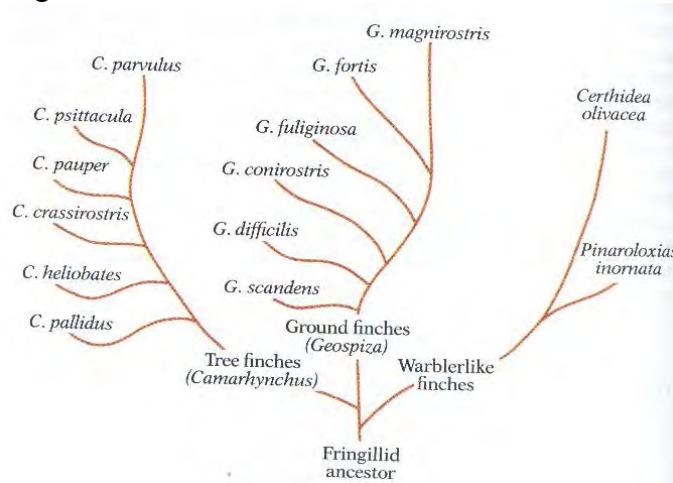
6. Which graph represents enzyme activity as a function of substrate concentration?



7. Which nitrogenous waste form is the least toxic, may be stored for long periods of time, and requires very little water for excretion?

- (A) Ammonia
- (B) Nitrogenous bases
- (C) Urea
- (D) Uric acid

8. The evolutionary history of the finches studied by Charles Darwin in the Galapagos Islands is shown in the diagram.



Ground finches feed on hard, large seeds. Tree finches feed on insects and small seeds. Warblers feed on insects.

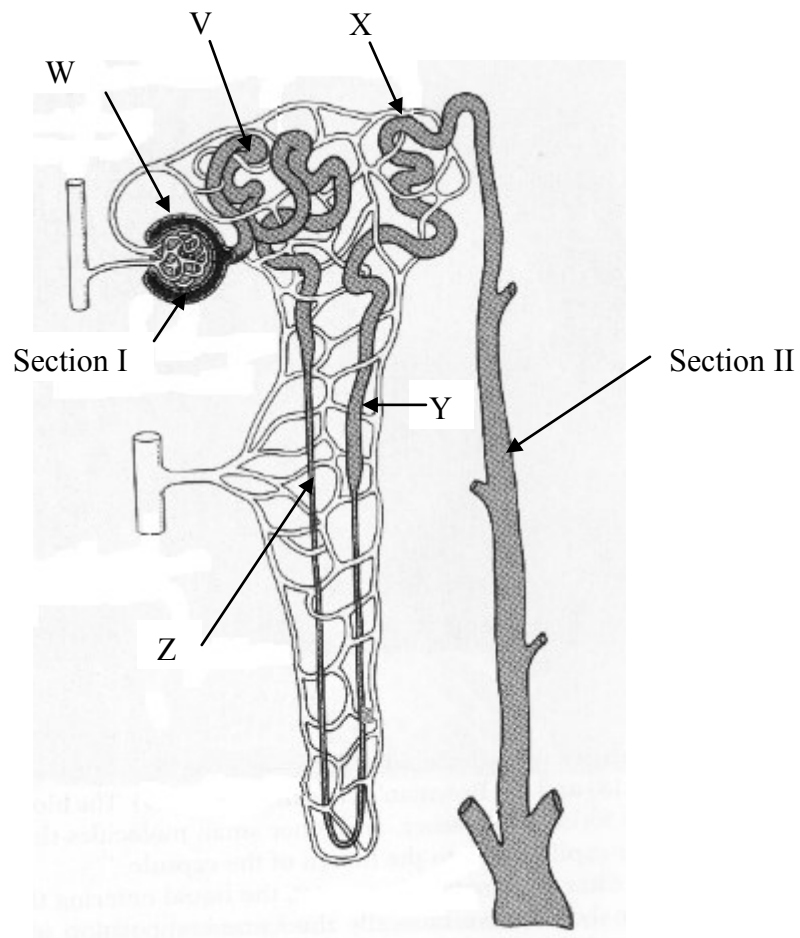
What type of evolution is shown by the finches?

- (A) Convergent evolution
 (B) Divergent evolution
 (C) Parallel evolution
 (D) Punctuated equilibrium
9. Antidiuretic hormone is affected by caffeine and stress: Caffeine inhibits the release of antidiuretic hormone from the pituitary gland, and stress increases the production of antidiuretic hormone. What would be the effects of caffeine and stress in terms of urine output and water level in blood?

	Urine output	Blood water levels
(A)	Caffeine: increases Stress: increases	Caffeine: increases Stress: increases
(B)	Caffeine: decreases Stress: increases	Caffeine: increases Stress: increases
(C)	Caffeine: increases Stress: decreases	Caffeine: decreases Stress: increases
(D)	Caffeine: decreases Stress: decreases	Caffeine: increases Stress: decreases

10. Blue Legged Crabs' haemoglobin equivalent — haemocyanin — does not bind to oxygen as efficiently when the crab is in a low potassium, low sodium environment. In order for the haemocyanin to function efficiently, the crab lowers its internal pH. What form of regulation is this an example of?
- (A) Active transport
 - (B) Enantiostasis
 - (C) Homeostasis
 - (D) Passive transport
11. What type of work did Thomas Boveri perform that helped our understanding of inheritance?
- (A) He studied the inheritance of genes on the X and Y chromosomes of fruit flies.
 - (B) He bred pea plants and studied the inheritance of one or two traits in the pea plants.
 - (C) He studied the inheritance of chromosomes in the daughter cells of horse worm egg cells undergoing meiosis.
 - (D) He studied red bread mould enzymes, showing that the loss of function in one enzyme led to the loss in function of one enzyme.
12. Coat colours of Andalusian fowls are an example of a trait determined by codominant alleles. Black, blue (a mixture of black and white) and white are the three phenotypes in Andalusian fowls. When blue fowls are crossed with white fowls, what percentage of the F_1 will be blue?
- (A) 0%
 - (B) 25%
 - (C) 50%
 - (D) 75%

13. The diagram is a cross-section of a mammalian nephron.



In which segment(s) does the reabsorption of water from the nephron via passive transport occur?

- (A) W and Section II
- (B) W, X, and Y
- (C) V, X, Y, Z and Section II
- (D) V, X, Z and Section II

14. What factors are important in the occurrence of an infectious disease besides the direct contact of individuals?
- (A) Air, water
 - (B) Water, chromosomes
 - (C) Air, water, food
 - (D) Air, water, food, vectors
15. What is the role of differentiation in maintaining health?
- (A) Results in different types of cells having different functions to perform.
 - (B) Enables certain cells to undergo rapid cell division.
 - (C) Controls the production of proteins called antibodies.
 - (D) Programming cell death.

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Exam Number

Write your Exam Number at the top of this Part A Answer Sheet.

Select the alternative A, B, C or D that best answers the question and fill in the response circle completely.

1. A ○ B ○ C ○ D ○
2. A ○ B ○ C ○ D ○
3. A ○ B ○ C ○ D ○
4. A ○ B ○ C ○ D ○
5. A ○ B ○ C ○ D ○
6. A ○ B ○ C ○ D ○
7. A ○ B ○ C ○ D ○
8. A ○ B ○ C ○ D ○
9. A ○ B ○ C ○ D ○
10. A ○ B ○ C ○ D ○
11. A ○ B ○ C ○ D ○
12. A ○ B ○ C ○ D ○
13. A ○ B ○ C ○ D ○
14. A ○ B ○ C ○ D ○
15. A ○ B ○ C ○ D ○

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Exam Number

Section I (continued)

Part B – 60 marks

Attempt Questions 16 - 30

Allow about 1 hour and 45 minutes for this part

Answer the questions in the spaces provided

Question 16 (3 marks)

(a) Identify a terrestrial Australian ectotherm. (1 mark)

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(b) Explain the responses of the ectotherm given in (a) to cold and hot environmental temperatures. (2 marks)

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Question 17 (7 marks)

Kidney failure is treated using a renal dialysis machine.

(a) Explain how metabolic wastes are removed via renal dialysis. (2 marks)

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Question 17 *Continued*

(b) Draw a longitudinal section of a mammalian kidney. Label two regions on your diagram. (3 marks)

(c) Compare the structures of the renal artery and renal vein. (2 marks)

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Question 18 (4 marks)

Unicellular organisms excrete dissolved nitrogenous wastes by the process of diffusion.

(a) Explain why the process of diffusion is inadequate for removing nitrogenous wastes from multicellular organisms. (2 marks)

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Questions continue on next page →

Question 18 *Continued*

(b) Outline the role of active transport in the mammalian kidney. (2 marks)

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Question 19 (5 marks)

The direction of movement of materials transported in the phloem tissue of plants is upwards and downwards from the leaves. Phloem tissue is composed of sieve tube cells and companion cells.

(a) Name a current theory for the movement of materials through phloem tissue. (1 mark)

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(b) Describe the theory you gave in (a). (4 marks)

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Question 20 (3 marks)

Carbon dioxide is one of the main metabolic waste products.

- (a) Describe a current technology that enables the measurement of carbon dioxide concentration in the blood. (1 mark)

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- (b) Justify the use of the technology you identified in (a). (2 marks)

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Question 21 (3 marks)

Aldosterone is a hormone secreted from the adrenal glands in humans. People who cannot secrete aldosterone suffer from Addison’s disease.

- (a) Describe the role of aldosterone in humans. (1 mark)

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Questions continue on next page →

Question 21 *Continued*

- (b) Explain how hormone replacement therapy is used to treat people that suffer from Addison's disease. (2 marks)

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Question 22 (4 marks)

In your course you presented a case study that demonstrated the impact of environmental change on the evolution of a species. Outline your case study below. (4 marks)

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Questions continue on next page →

Question 24 (5 marks)

Natural selection is one mechanism that accounts for Darwin/Wallace's theory of evolution.

- (a) Using a modern example, explain how natural selection contributes towards the evolution of an organism. (3 marks)

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- (b) Describe the potential role that mutations may have on evolution. (2 marks)

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Questions continue on next page →

Question 26 (3 marks)

- (a) Using the symbols provided in the key, construct a pedigree for the inheritance of red hair colour in the Richards family described.

Red hair colour in the Richards family

Mrs Richards is a brown-haired female married to Mr Jones, a brown-haired male. They have three children, a male (Jack) who is red-haired, a male (Mark) who is brown-haired and a female (Jill) who is red-haired. Jack marries a black-haired female and they have one red-haired female, one black-haired female and one red-haired male. Jill marries a brown-haired male and they have three children, two males and a female. All of Jill's children are brown-haired.

Key

Red-haired male



brown or black-haired male



Red-haired female



brown or black-haired female

- (b) State the type of genetic inheritance for red hair colour in the family. (1 mark)
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Question 27 (5 marks)

The treatment of water for human consumption is an important process for the prevention and control of disease.

- (a) Describe two ways by which water is treated for human drinking. (2 marks)

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- (b) Describe how you could identify if microbes are present in water sample. (3 marks)

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Questions continue on next page →

Question 28 (2 marks)

The table shows the proportions of nucleotide bases in DNA from a variety of organisms.

organism	adenine	thymine	cytosine	guanine
bacterium	1.1	1.0	2.6	2.6
calf thymus	1.7	1.6	1.0	1.0
yeast	1.8	1.9	1.0	1.0

Explain the significance of these results with respect to the work of James Watson and Francis Crick in the 1950s.

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Questions continue on next page →

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Section II

Student Number

25 marks**Attempt ALL parts of Question 33 Genetics-The Code is Broken?****Allow about 45 minutes for this part**

Questions continue on next page →

Answer the question parts in a writing booklet. Extra writing booklets are available.

Question 33 Genetics – The Code is Broken? (25 marks)

- (a) Compare the inheritance of a multiple allele trait with a trait that follows Mendelian inheritance. (2 marks)
- (b) Outline the history of the selective breeding of an agricultural species and explain the purpose of the selective breeding of this species. (4 marks)
- (c) Compare the processes and products of gene cloning with whole organism cloning. (6 marks)
- (d) Describe the role of gene cascades in determining limb formation in birds and mammals. (2 marks)
- (e) (i) Name a disease that may be treated by gene therapy. (1 mark)
- (ii) Describe how gene therapy may be used to treat the disease you named in (i). (2 marks)
- (f) (i) Give one example of a mutation that is a result of chromosomal rearrangements (not a change in chromosomal number). (1 mark)
- (ii) Describe how this mutation may occur. (1 mark)
- (iii) Name one genetic mutation that affects human health. (1 mark)
- (iv) Describe the effect of the genetic mutation you gave in (iii) on human health. (1 mark)

- (g) A heterozygous red-bodied, long-winged insect (RrLl) is crossed with a white-bodied, short-winged fruit fly (rrll). Numbers of F₁ offspring produced are displayed in the table.

F ₁ offspring phenotype	number of offspring
heterozygous red-bodied, short-winged	75
white-bodied, heterozygous long-winged	77
heterozygous red-bodied, heterozygous long-winged	493
white-bodied, short-winged	500

- (i) State the name of this type of cross, which was designed to show the relative positions of genes along a particular chromosome. (1 mark)
- (ii) Explain how the outcomes of the cross indicate the two genes involved are linked. (2 marks)
- (iii) Describe how these results may be used to calculate the map distance between the two genes. (1 mark)

End of Trial paper

James Ruse Agricultural High School
2008 TRIAL HIGHER SCHOOL CERTIFICATE EXAMINATION
BIOLOGY MARKING GUIDELINES

PART A

Multiple Choice: Questions 1-15 (1 mark each)

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|-----|------------------------------------|------------------------------------|------------------------------------|------------------------------------|
| 1. | A <input type="radio"/> | B <input type="radio"/> | C <input checked="" type="radio"/> | D <input type="radio"/> |
| 2. | A <input type="radio"/> | B <input checked="" type="radio"/> | C <input type="radio"/> | D <input type="radio"/> |
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| 4. | A <input type="radio"/> | B <input type="radio"/> | C <input checked="" type="radio"/> | D <input type="radio"/> |
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| 9. | A <input type="radio"/> | B <input type="radio"/> | C <input checked="" type="radio"/> | D <input type="radio"/> |
| 10. | A <input type="radio"/> | B <input checked="" type="radio"/> | C <input type="radio"/> | D <input type="radio"/> |
| 11. | A <input type="radio"/> | B <input type="radio"/> | C <input checked="" type="radio"/> | D <input type="radio"/> |
| 12. | A <input type="radio"/> | B <input type="radio"/> | C <input checked="" type="radio"/> | D <input type="radio"/> |
| 13. | A <input type="radio"/> | B <input type="radio"/> | C <input type="radio"/> | D <input checked="" type="radio"/> |
| 14. | A <input type="radio"/> | B <input type="radio"/> | C <input type="radio"/> | D <input checked="" type="radio"/> |
| 15. | A <input checked="" type="radio"/> | B <input type="radio"/> | C <input type="radio"/> | D <input type="radio"/> |

PART B

Total marks: 60 marks

Question 16 (3 marks)

(a) Gecko

(1): Stated name of an Australian ectotherm.

(b) Cold temperature: gecko seeks sun by basking; this enables the gecko to warm up. The gecko's body temperature depends on the environment.

(1): body temperature of gecko depends on the environment.

(1): correct responses given.

Question 17 (7 marks)

(a) Wastes such as urea are removed via diffusion from blood into the dialysis solution. This occurs via the blood passing through dialysis tubing which has a semipermeable membrane.

(1): diffusion

(1): dialysis tubing has a semipermeable membrane

(b) labelling any two of: cortex, medulla, pelvic region.

(c) Renal artery has a smaller diameter, thicker muscular walls, and more elastic tissue than the renal vein. Veins contain valves.

(1): one difference between arteries and veins.

(1): one difference between arteries and veins.

Question 18 (4 marks)

(a) Multicellular organisms have many cells which produce nitrogenous wastes generally in the form of ammonia. Diffusion is inadequate in this case since there is too much nitrogenous waste to be excreted via this method. Diffusion is not as efficient as a kidney in ridding the body of nitrogenous waste since the cells form tissues and organs, and nitrogenous waste would simply diffuse from one cell to another.

(1): excessive nitrogenous waste for diffusion.

(1): diffusion is inefficient due to the structure of cells forming tissues and organs.

(b) Active transport is required to move substances across cell membranes usually against a concentration gradient. This process enables the reabsorption of nutrients such as glucose and phosphate ions from the proximal tubule into the blood. Active transport also allows the secretion of large organic substances from the blood into the descending loop of Henle.

(1): Two of the above given.

(1): Only one of the above given.

Question 19 (5 marks)

(a) Pressure-flow hypothesis; source-to-sink theory.

(1): name of theory given.

(b) The leaves and other photosynthetic regions of the plant make glucose via photosynthesis. The glucose is actively transported to the phloem. In the phloem, the glucose attracts water via osmosis, resulting in a high pressure. The glucose solution then moves to areas that have lower pressure (the sinks). At these areas the cells actively transport the glucose and use it for cellular respiration. Water then follows the glucose in the cells via osmosis.

(1): active transport of glucose at source to phloem

(1): pressure gradient occurs

(1): sink cells actively transport sugar

(1): role of osmosis

Question 20 (5 marks)

(a) Blood gas analysis machine (arterial blood gas analysis).

(b) Carbon dioxide reacts with water to form carbonic acid, which makes the cellular environment acidic. The functioning of enzymes in cells is affected by the acidity. Therefore the levels of carbon dioxide must be monitored.

(1): carbonic acid formation.

(1): enzymes are affected by the pH.

Question 21 (3 marks)

(a) Reabsorption of sodium in the distal convoluted tubule of nephrons. (1)

(b) Name of synthesised hormone: Florinef, fludrocortisone. (1)

Administration of steroids via oral or injection on a regular basis. (1)

Question 22 (4 marks)

Early kangaroos: lived in a cool, heavily forested environment; being small enabled the kangaroos (such as the Musky-rat kangaroo) to move around easily to obtain food; teeth would have been adapted for browsing (chewing leaves and stems).

About 10-12 million years ago, Australia was much drier (due to the drift northwards) and the kangaroos that survived the drier environment were those that could hop (fossil record) and had teeth for either browsing or grazing (eating grass). These adaptations would become more important as the conditions got drier (end of the Pliocene).

It is thought that an early tertiary ancestor of the Kangaroo family browsed on leaves and shoots of shrubs. As the continent became drier, the grazing lifestyle may have developed. Studies of the teeth of kangaroos of different genera show that kangaroos (e.g. swamp wallaby) whose food contains little abrasive material such as tough grass have a different tooth and jaw structure compared to those kangaroos that graze (e.g. grey kangaroo)

(1): one change described for a species

(1): change is linked to environmental selection pressures

(1): a second change described for the species

(1): change is linked to environmental selection pressures

Question 23 (4 marks)

- Palaeontological evidence refers to the fossil record and this provides support for the theory of evolution. For example, the modern horse, *Equus*, has had several ancestors, as found in the fossil record.
- These horse fossils show gradual changes over the course of some 65 million years.
- The earliest horse fossil for instance, *Hyracotherium*, had a three-toe foot structure whilst the younger horse fossil, *Miohippus*, had evolved to have mainly a single-toe foot with two side bones; the *Equus* has a single toe foot. These changes are thought to be the result of the horse evolving in an environment that was changing from marsh-like to one that was dry and of hard ground.

(1): relevant example of a fossil stated. (ex)

(1): description of how the fossil record supports the theory of evolution. (d)

(1): elaboration of the description. (f)

(1): demonstrated understanding of evolution.

- Only three of the above points given.(3)
- Only two of the above points given. (2)
- Only one of the above points given. (1)

Question 24 (5 marks)

(a) Peppered moth in England: Moth populations in England in the mid 1800s consisted mainly of light-coloured moths with a small number of dark-coloured moths. However, with the advent of the industrial revolution, the trunks of forest trees became darker as a result of the accumulation of soot from the factories. The darker colour caused the lighter coloured moths to become conspicuous to preying birds and their numbers declined. The dark coloured moths increased in frequency due to their cryptic advantage. Thus, the change in the environment favoured the darker moths and the moth population contained a greater proportion of dark coloured moths than previously.

(1): modern example given

(1): environmental change effect on the survival of the organism

(1): impact of environment on the numbers of the organism and hence evolution

(b) Mutations refer to a change in the DNA structure or numbers of chromosomes in cells. Mutations provide a source of genetic variation that evolution may act upon. This variation may be advantageous for the survival of organisms.

(1): description of mutation

(1): impact of mutations on evolution

Question 25 (5 marks)

Investigated the impact of UV light on the growth of radishes.

(1): 'control' condition: radish seedlings in the dark (cupboard). (c)

(1): 'experimental' condition: radish seedlings exposed to UV light. (e)

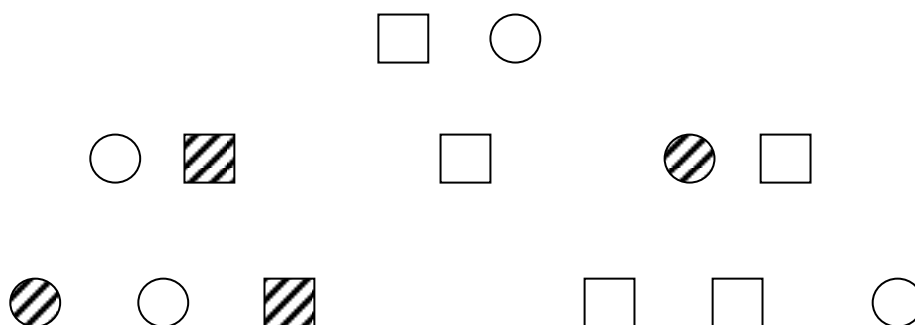
(1): controlling variables: same type of soil used, same amounts of water, same type of radish seeds used in each punnet, maintained at the same temperature. Must have two variables controlled or replicates described. (v)

(1): growth was determined by measuring the height of seedlings: from edge of punnet to top of all seedlings (average results).

(1): results observed – radishes grown in dark did not grow as high as those growing exposed to UV light. Radishes in the dark had yellow-coloured leaves whereas those grown in UV light had green-coloured leaves.

Question 26 (3 marks)

(a)



(1): Pedigree: correct offspring shown

(1): correct symbols used

(b) autosomal recessive. (1)

Question 27 (5 marks)

(a) Micro-filter water to remove undissolved solids and cells; add a flocculating agent to trap small particles and bacteria, filter this away; add chlorine to water in order to destroy bacteria and any other organism; boil water.

Any two of the above.

(b) Aseptically apply an inoculating loop of the water sample to a sterile agar plate. Incubate the plate and observe for colonies of microbes. Compare with control (no water) agar plate.

(1): aseptic techniques described

(1): sterile agar used as growth medium

(1): observe for growth of colonies on agar after incubation

Question 28 (2 marks)

The results show that the abundance of nitrogenous bases corresponds to specific pairing (A-T, G-C). This implied that the nitrogenous must be associated (bonded) in the DNA structure and verified that DNA is a double helix structure.

(1): specific pairing

(1): DNA structure is a double helix

Question 29 (7 marks)

Animals:

This method involves the direct microinjection of a chosen gene construct (a single gene or a combination of genes) from another member of the same species or from a different species, into the pronucleus of a fertilized ovum. (m)

The DNA construct (usually about 100 to 200 copies in 2 pl of buffer) is introduced by microinjection through a fine glass needle into the male pronucleus - the nucleus provided by the sperm before fusion with the nucleus of the egg.

The insertion of DNA is, however, a random process, and there is a high probability that the introduced gene will not insert itself into a site on the host DNA that will permit its expression. The manipulated fertilized ovum is transferred into the oviduct of a recipient female, or foster mother that has been induced to act as a recipient by mating with a vasectomized male.

This method has been successful in producing organisms such as transgenic sheep that produce blood clotting proteins in their milk.

(1): microinjection

(1): pronucleus of fertilised ovum

(1): ovum placed into recipient female

(1): one use of transgenic animals

Plants:

Use of a bacterium such as *Bacillus thuringiensis* (Bt) that has a foreign gene inserted into a plasmid. The bacterium then inserts plasmids into the root cells of a plant.

Many plants have been successfully genetically modified using this method. For example, cotton has been inserted with a gene that produces a Bt toxin that is an insecticide against moths, which is a pest of cotton crops.

(1): use of a bacterium

(1): foreign gene inserted into bacterial plasmid

(1): one use of transgenic plants

Question 33 Genetics – The Code is Broken? (25 marks)

(a) Multiple allele traits are those that have three or more alleles for a gene. Multiple allele traits have a greater number of genotypes and phenotypes than Mendelian traits, which only are based upon the inheritance of two alleles.

(1): definitions of multiple allele traits and Mendelian traits

(1): multiple allele traits exhibit greater number of genotypes and phenotypes

(b)

(1): selective breeding reason

(1): named species.

(1): one major historical breeding event described.

(1): one other major historical breeding event described.

(-1 if not used for agricultural purposes)

Selective breeding: the purposeful selection of two organisms for breeding in order to produce offspring that have desirable characteristics.

(c)

Criteria	Marks	Mark code
Gene cloning requires the use of <u>recombinant DNA technology</u> (1) techniques in order to <u>replicate the gene via the reproduction of an organism</u> (1) that carries the gene (e.g., the plasmids in bacterial cells). This process <u>yields lots of genes</u> that are <u>exactly the same</u> (1) as the original gene.	1	R
	1	B
	1	G
Whole organism cloning involves the <u>nucleus of a donor cell</u> being placed, via a micropipette, into an <u>enucleated egg cell</u> . (1) This <u>cell is placed into a surrogate mother</u> (1) who produces an organism that is a replica of the donor. This process yields an <u>organism that has the same genome as the donor cell provider</u> . (1)	1	M
	1	S
	1	O

(d)

Criteria	Marks	Mark code
Gene cascades also play a role in the development of the embryo. If one of the transcription factor proteins switches on another gene that also is a transcription factor, the latter may switch on another gene(s), which may also be responsible for transcription. (1) In this fashion a cascade of gene expression is built up (a type of amplification), producing proteins along the way that may influence what the embryonic cells differentiate into. (1)	1	T
	1	D

(e) (i) Cystic fibrosis (1)

(ii) Cystic fibrosis is a result of a defective cell membrane protein in the some lung tissues. This is a result of a mutated or a missing gene for the membrane protein. To treat this condition via gene therapy, the normal gene for the membrane protein is inserted into a viral vector. (1) This viral vector is inserted into the lung tissue via a tube inserted in the patient's nose, leading directly into the lungs. (1)

(f) (i) Translocation Down syndrome; cri-du-chat syndrome (1)

(ii) Translocation Down syndrome: part of chromosome 21 attaches to chromosome 14.
Cri-du-chat syndrome: missing part of chromosome 5.

(1): description of the chromosomal arrangement.

(iii) Cystic fibrosis; sickle cell anaemia

Cystic fibrosis: excessive secretions of mucous that block airways and inhibit pancreas secretions; these may lead to lung infections.

Sickle-cell anaemia: fatigue, breathlessness upon physical exertion.

(g) (i) test cross (1)

(ii) The frequencies of the two types of recombinants (1) are similar and lower than the parental frequencies (1).

(iii) The total of the frequencies of the two types of recombinants is divided by the total number of offspring. (1)