Insight Publications

INSIGHT

Trial Exam Paper

2007
BIOLOGY
Written examination 2

Solutions book

This book presents:

- correct solutions
- explanatory notes
- mark allocations

SECTION A – Multiple-choice questions

Question 1

In humans, gametes are cells which pass on genetic information from parent to offspring. Gametes are normally haploid and arise from germ line cells. Germ line cells are

A. haploid.

B. **diploid.**

C. tetraploid.

D. polyploid.

Answer is B

Explanatory notes

- A is incorrect because only gametes (eggs and sperm) are haploid.
- C is incorrect because a tetraploid cell would have four times the haploid number (4n) of a germ line cell.
- D is incorrect because a polyploid cell has three or more times the haploid number of a germ line cell.

Question 2

The chromosomes in a human somatic cell can be arranged to form a karyotype. In a normal human male the chromosomes in a karyotype will be arranged into

A. 46 pairs of chromosomes, which include one large X chromosome and a smaller Y chromosome.

B. 44 pairs of chromosomes, which include one large X chromosome and a smaller Y chromosome.

C. 23 pairs of homologous chromosomes, including one large X chromosome and a smaller Y chromosome.

D. **22 pairs of homologous chromosomes and one large X chromosome and a smaller Y chromosome.**

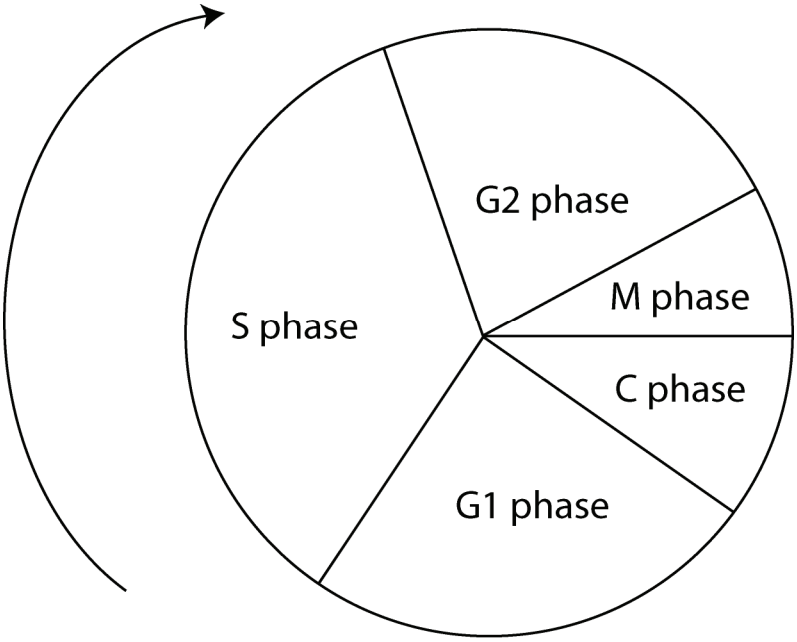
Answer is D

Explanatory notes

- A is incorrect because a normal human somatic cell has a diploid number of 46 not 92.
- B is incorrect because a normal human somatic cell has a diploid number of 46 not 88.
- C is incorrect because there are 22 pairs of homologous (matched) autosomes and 1 pair of unmatched sex chromosomes.

Question 3

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.



It would be expected that the mass of the cell would increase measurably during

A. **S phase.**

B. G1 phase.

C. G2 phase.

D. M and C phases.

Answer is A

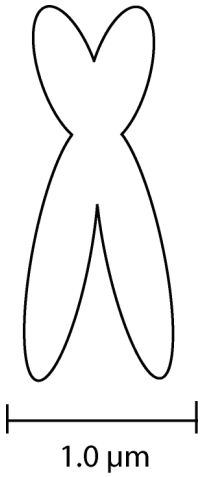
Explanatory notes

- B is incorrect because even though the cell is growing in G1 phase, it has not yet replicated its DNA and therefore its mass will not yet have increased.
- C is incorrect because the cell has already replicated its DNA and increased in mass in S phase – the synthesis phase.
- D is incorrect because the cell has already replicated its DNA and increased in mass in S phase.

Use the following information to answer Questions 4 and 5.

Question 4

When a eukaryote cell prepares to divide it, duplicates each of its chromosomes. The diagram shows a chromosome after duplication.



From the diagram, a duplicated chromosome consists of

A. a double-stranded chromosome which narrows at the centriole.

B. an homologous pair of chromosomes which narrow at the centromere.

C. **two sister chromatids which narrow at the centromere.**

D. two sister chromatids which narrow at the centriole.

Answer is C

Explanatory notes

- A is incorrect because even though the diagram shows a double-stranded chromosome, it narrows at the centromere, not the centriole. The centriole is a cylindrical organelle of microtubules which functions in the organisation of the spindle fibres during mitosis and meiosis.
- B is incorrect because the diagram does not depict an homologous pair of chromosomes.
- D is incorrect because even though the diagram shows two sister chromatids, they do not narrow at the centriole.

Question 5

A duplicated chromosome viewed through a scanning electron microscope (SEM) shows a hairy appearance because each strand consists of a very long fibre which is folded and coiled in a compact arrangement. This fibre is

A. **a DNA-protein complex known as chromatin.**

B. also known as a nucleosome.

C. made up entirely of histones.

D. part of the troponin complex.

Answer is A

Explanatory notes

- B is incorrect because a nucleosome is a section of DNA molecule which loops twice around a core of eight histone proteins.
- C is incorrect because a chromosome consists of chromatin (DNA and histone proteins), not just histones.
- D is incorrect because the troponin complex is a set of regulatory proteins associated with control of muscle contraction.

Question 6

In living organisms genes are regarded as the smallest physical unit of heredity. Different genes have

A. the same length open reading frame.

B. **different nucleotide sequences.**

C. the same number of nucleotides.

D. identical nucleotide sequences.

Answer is B

Explanatory notes

- A is incorrect because the open reading frame for genes can be of differing lengths depending on the gene.
- C is incorrect because different genes have different numbers of nucleotides in each nucleotide sequence.
- D is incorrect because different genes cannot have identical nucleotide sequences otherwise they would be the same genes.

Question 7

Genes contain coded instructions for joining specific amino acids into proteins. Each segment of the coding region of a gene is known as the

A. **exon.**

B. intron.

C. promoter.

D. flanking region.

Answer is A

Explanatory notes

- B is incorrect because the intron is the non-coding part of the gene.
- C is incorrect because the promoter is the nucleotide sequence in DNA that binds RNA polymerase and indicates where to begin transcribing RNA.
- D is incorrect because flanking regions are the regions located either upstream or downstream of the coding region of a gene.

Question 8

After mating, two polled (hornless) cattle produced a calf with horns. It would be reasonable to conclude that

A. **all horned × horned matings would produce only horned offspring.**

B. all polled × polled matings would produce horned offspring.

C. the presence or absence of horns is a monogenic trait with three alleles.

D. horned is dominant to polled.

Answer is A

Explanatory notes

- A is correct because the genotype of hornless cows could be **Hh**, which when crossed would produce 50% **Hh**, 25% **hh** and 25% **HH** – corresponding with 75% hornless (polled) and 25% horned.
- B is incorrect because there is not enough evidence provided to support the idea that only horned offspring will be produced.
- C is incorrect because there is not enough evidence to support the existence of four different phenotypes.
- D is incorrect because if the horned trait was dominant, two polled cows would not be able to produce a horned calf.

Use the following information to answer Questions 9 and 10.

Question 9

In guinea pigs, two genes and their alleles are

Gene for coat length	**L**	short
	l	long

Gene for coat colour	**B**	black
	b	white

These genes control two unrelated characteristics and therefore

A. are considered to be linked.

B. do not assort independently.

C. are found on homologous chromosomes.

D. **assort independently.**

Answer is D

Explanatory notes

- A is incorrect because there is no linkage between unrelated characteristics (genes).
- B is incorrect because unrelated characteristics determined by unlinked genes assort independently.
- C is incorrect because if the genes are found on an homologous pair of chromosomes they are linked.

Question 10

Guinea pigs with the genotype **LlBb** will have

A. long, white-haired coats.

B. long, black-haired coats.

C. short, white-haired coats.

D. **short, black-haired coats.**

Answer is D

Explanatory notes

- A is incorrect because the genotype for long, white-haired coats is **llbb**.
- B is incorrect because the genotype for long, black-haired coats is **llBb or llBB**.
- C is incorrect because the genotype for short, white-haired coats is **LLbb or Llbb**.

Question 11

An anticodon is a triplet sequence of bases found on a

A. mRNA molecule.

B. template strand of DNA.

C. **tRNA molecule.**

D. rRNA molecule.

Answer is C

Explanatory notes

- A is incorrect because the triplet found on a mRNA molecule is called a codon.
- B is incorrect because the triplet sequence found on template DNA is referred to as a DNA triplet.
- D is incorrect because bases found on rRNA are not referred to as anticodons.

Use the following information to answer Questions 12 and 13.

The sequence of bases in the template strand of a piece of DNA is

A A G T C C A T G G A C

Question 12

The sequence of bases in the complementary strand of the same piece of DNA would be

A. A A G T C C A T G G A C

B. C C T G A A C G T T C A

C. U U C A G G U A C C U G

D. **T T C A G G T A C C T G**

Answer is D

Explanatory notes

- A is incorrect because this is not the correct complementary sequence (A should pair with T; C with G).
- B is incorrect because this is not the correct complementary sequence (A should pair with T; C with G).
- C is incorrect because uracil is not a nucleotide in DNA.

Question 13

The base sequence of the mRNA produced from this piece of DNA would be

A. T T C A G G T A C C T G
B. **U U C A G G U A C C U G**
C. A A G U C C A U G G A C
D. A A G T C C A T G G A C

Answer is B

Explanatory notes

- A is incorrect because a sequence of mRNA should contain uracil (U) not thymine (T).
- C is incorrect because the sequence of mRNA is not complementary (A – U; C – G).
- D is incorrect because the sequence of mRNA is not complementary nor does it contain uracil.

Question 14

During embryonic development there are active genes that control the expression of structural genes thus ensuring that body parts are built in the correct positions. These genes are known as

A. **homeotic genes.**
B. structural genes.
C. regulator genes.
D. functioning genes.

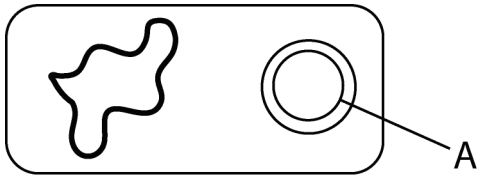
Answer is A

Explanatory notes

- B is incorrect because structural genes are controlled by homeotic genes, not themselves.
- C is incorrect because regulator genes determine whether other genes are active or not.
- D is incorrect because functioning genes describes a gene which is operating as it should be.

Question 15

The diagram represents a bacterial cell. The structure labelled **A** found in many bacterial cells is known as a



A. primer.
B. promoter.
C. probe.
D. **plasmid.**

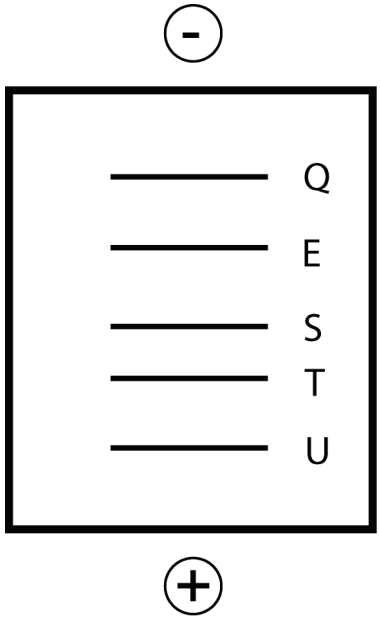
Answer is D

Explanatory notes

- A is incorrect because a primer is a short, single-stranded DNA or RNA sequence complementary to a DNA fragment.
- B is incorrect because a promoter is part of the upstream flanking region which contains base sequences that control the activity of the gene.
- C is incorrect because a probe is a fluorescent or radioactive single-stranded sequence of DNA or RNA with a base sequence complementary to a sequence in a target strand of DNA.

Use the following information to answer Questions 16 and 17.

A segment of linear DNA was isolated and cut using a restriction enzyme. The fragments of DNA were then placed in a gel and separated by electrophoresis. The diagram below shows the gel and fragments at the end of electrophoresis.



Question 16

Prior to cutting with the restriction enzyme, the segment of DNA has

A. eight recognition sites.
B. five recognition sites.
C. **four recognition sites.**
D. three recognition sites.

Answer is C

Explanatory notes

- A is incorrect because if there were eight recognition sites there would be nine bands of DNA in the gel.
- B is incorrect because if there were five recognition sites there would be six bands of DNA in the gel.
- D is incorrect because if there were three recognition sites there would be four bands of DNA in the gel.

Question 17

Of the fragments obtained in the gel, which is the largest fragment?

A. **Fragment Q is the largest.**
B. Fragment S is the largest.
C. Fragment U is the largest.
D. All fragments are the same size.

Answer is A

Explanatory notes

- Gel electrophoresis can separate molecules on the basis of size and charge. Since DNA molecules carry a net negative charge in solution, they will always migrate to the positively charged electrode. Smaller DNA fragments move more easily through agarose gel and so travel further compared with larger DNA fragments.
- B is incorrect because Fragment S is halfway down the gel, thus it is smaller than Fragments Q and R.
- C is incorrect because Fragment U is at the end of the gel, indicating it is the smallest piece.
- D is incorrect because the bands appear at five distinct positions in the gel, indicating they are all of different sizes.

Question 18

Seed mass is a phenotype shown in bean plants. It has been demonstrated that on occasions heavy parental (P) seeds can produce F1 plants with seeds of lighter mass than normal. In addition these lighter seeds demonstrate a range of masses. This can be explained by the fact that seed mass is an example of

A. genetic drift.
B. discontinuous variation.
C. **polygenic inheritance.**
D. monogenic inheritance.

Answer is C

Explanatory notes

- A is incorrect because genetic drift describes changes in allele frequency due to chance alone and not natural selection, there is no data to support this.
- B is incorrect because discontinuous variation occurs when a population can be grouped into a few non-overlapping classes with respect to phenotype; this is not evident in the example.
- D is incorrect because monogenic traits show discontinuous variation in a population; the example indicates a phenotype which shows continuous variation.

Use the following information to answer Questions 19 and 20.

In shorthorn cattle, coat colour is controlled by a gene with the alleles **R** (red hair) and **r** (white hair). A heterozygous cow will have a roan coat (mixture of both red hairs and white hairs). A population of one hundred cows was sampled to investigate coat colour. The results are shown in the table.

genotype based on coat colour	**RR**	**Rr**	**rr**
number of cows of each genotype	80	10	10

Question 19

The relationship that best describes this expression of alleles is

A. complete dominance.

B. incomplete dominance.

C. **co-dominance.**

D. partial dominance.

Answer is C

Explanatory notes

- A is incorrect because with complete dominance, the phenotypes of the heterozygote and dominant homozygote are indistinguishable and this is not the case in shorthorn cattle.
- B is incorrect because incomplete dominance occurs when one trait is not fully dominant over its partner, resulting in an intermediate phenotype in the heterozygote.
- D is incorrect because partial dominance occurs when one trait is not fully dominant over its partner, resulting in an intermediate phenotype in the heterozygote.

Question 20

From the data above, it is possible to conclude that

A. there is a total of 100 alleles in this population.

B. the frequency of the **R** allele is 0.9.

C. the frequency of the **r** allele is 0.3.

D. **the total number of r alleles is 30.**

Answer is D

Explanatory notes

- A is incorrect because there is a total of 200 alleles in this population (each individual has 2 alleles).
- B is incorrect because the freq(**R**) = the number of **R** alleles/total number of alleles = 0.85
- C is incorrect because the freq(**r**) = the number of **r** alleles/total number of alleles = 0.15

Question 21

In a population of living organisms, allele frequencies can change over time. The change in allele frequencies is **not** due to

A. **random mating.**

B. chance.

C. migration.

D. selection.

Answer is A

Explanatory notes

- B is incorrect because chance events lead to random genetic drift which changes allele frequencies.
- C is incorrect because migration (gene flow) does change the allele frequencies of a population.
- D is incorrect because selection (competition for resources) leads to a change in allele frequencies.

Question 22

Mitochondrial DNA (mtDNA) can be used to trace the evolutionary history of a species. A characteristic of mtDNA **not** used to distinguish it from nuclear DNA is

A. lack of recombination.

B. descent through the maternal line.

C. high copy number.

D. **molecular structure.**

Answer is D

Explanatory notes

- A is incorrect because the lack of recombination in mtDNA does distinguish it from nuclear DNA.
- B is incorrect because nuclear DNA is inherited through both the maternal and paternal lines.
- C is incorrect because each mitochondrion carries up to 10 mtDNA molecules with each cell carrying several hundred mitochondria. In the case of nuclear DNA, only 2 copies of each chromosome are present in somatic cells.
- D is correct because the molecular structure of mtDNA and nuclear DNA are identical and therefore cannot be used to distinguish one from the other.

Question 23

Carbon-14 (^{14}C) dating is used to estimate the age of fossilised organic material up to the age of 60 000 years. In comparison, electron spin resonance (ESR)

A. relies on radioactive decay in order to make its estimation of fossil age.

B. is used to estimate the age of material more than one million years old.

C. determines the alignment of the Earth's magnetic field at the time when the sample was last heated above a critical level.

D. **measures the microwave energy absorbed by samples exposed to radiant energy in the distant past.**

Answer is D

Explanatory notes

- A is incorrect because ESR does not rely on radioactive decay; it is a non-isotopic method of dating.
- B is incorrect because ESR is used to estimate the age of material from 50 000–500 000 years old.
- C is incorrect because palaeomagnetism determines the alignment of the Earth's magnetic field, not ESR.

Question 24

The evolution of bipedal locomotion in early hominins was accompanied by significant modifications in the skeleton. One of the characteristics **not** associated with bipedal locomotion is the

A. **shape and positioning of the shoulder blades.**

B. arrangement of the femur and tibia.

C. structure and shape of the pelvis and hip bone.

D. position of the foramen magnum.

Answer is A

Explanatory notes

- B is incorrect because the outward slant of the femur and tibia indicates presence of bipedalism.
- C is incorrect because a bowl-shaped pelvis and short hip bone is indicative of bipedalism.
- D is incorrect because a forward-positioned foramen magnum is consistent with bipedalism.

Question 25

The origin of the human species is thought to have been in Africa. Rather than evolving in a simple linear progression, the hominin line has been shown to have emerged in a branching manner with several different hominin species coexisting. Despite this, it has been determined that the hominin species appeared in the fossil record from oldest to most recent in the following order

A. *A. anamensis, A. africanus, H. sapiens, H. habilis*.

B. ***A. afarensis, A. africanus, H. habilis, H. heidelbergensis.***

C. *H. sapiens, H. habilis, A. africanus, A. ramidus*.

D. *A. ramidus, H. rudolfensis, A. africanus, H. heidelbergensis*.

Answer is B

Explanatory notes

- A is incorrect because *H. sapiens* occurs in the evolution of humans after the appearance of *H. habilis*, not before.
- C is incorrect because the hominins in the genus *Homo* (*H. sapiens, H. habilis*) appear in the evolutionary tree after the appearance of Australopithecine *(A. africanus*) and Ardipithecines (*A. ramidus)*, not before them.
- D is incorrect because the order of appearance of species in the evolutionary tree is *A. ramidus, A. africanus, H. rudolfensis, H. heidelbergensis*, not the order listed in D.

END OF SECTION A

SECTION B – Short-answer questions

Question 1

In cats the ability to produce a tail is genetically controlled by a single gene. The gene which controls the development of a tail in Manx cats has the following alleles:

$\mathbf{M}^{L}$ – taillessness **M** – normal tail

It is known that tailless Manx cats are heterozygous ($\mathbf{M}^{L}\mathbf{M}$) and carry a recessive allele for normal tail.

1a. What would be the genotype of a cat with a normal tail?

Solution

M M

1 mark

Explanatory notes

- The allele for normal tail is **M**, therefore the genotype (which consists of 2 alleles) would be **M M**.

A cross is made between a Manx cat and a cat with a normal tail.

1b. What genotypic ratio and phenotypic ratio would be expected in the offspring of the cross between these two cats? Ensure that the correct allelic symbols are used.

Solution

Parental cross:	$\mathbf{M}^{L}\mathbf{M} \times \mathbf{M}\ \mathbf{M}$	
Offspring:	½ $\mathbf{M}^{L}\mathbf{M}$: ½ **M M**	(genotypic ratio)
Phenotype:	1 Manx : 1 normal tail	(phenotypic ratio)

3 marks

Explanatory notes

- Students should provide the answers set out in the manner demonstrated. Correct allelic symbols must be used and the genotypic and phenotypic ratios must be clearly indicated.

Mark allocation

- 1 mark – correct allelic symbols
- 1 mark – correct genotypic ratios
- 1 mark – correct phenotypic ratios

A research group studying inheritance patterns performed a large number of crosses with the Manx offspring from the initial cross. They found that their crosses always produced offspring in a ratio of two Manx cats to one normal tailed cat (2 Manx: 1 normal)

1c. What is the most likely explanation for this result?

Solution

The genotype $\mathbf{M^LM^L}$ is a lethal genotype and any zygotes formed with this genotype will not develop or survive to birth.

1 mark

Explanatory notes

- Possible genotypes from this cross are $\mathbf{M^LM^L}$, $\mathbf{M^LM}$ (Manx) and **MM** (normal). The fact that the genotype $\mathbf{M^LM^L}$ never appears, despite the numbers of crosses, suggests that it is lethal or kills cats with this genotype.

Huntington's disease is a degenerative medical condition which affects adult humans, usually between the ages of 35–45 years. It is characterised by deterioration of the nervous system and is fatal.

1d. Why does Huntington's disease persist in the human population if it is fatal?

Solution

Huntington's disease may not necessarily take effect until after the onset of adulthood; consequently, the lethal allele can be passed on to offspring before the condition emerges in parent(s).

2 marks

Explanatory notes

- Even though Huntington's disease is fatal (caused by lethal alleles) it may not necessarily take effect until after the onset of adulthood and more significantly, after reproductive maturity has been reached. Thus the allele can be passed on to children before it has any phenotypic effect.

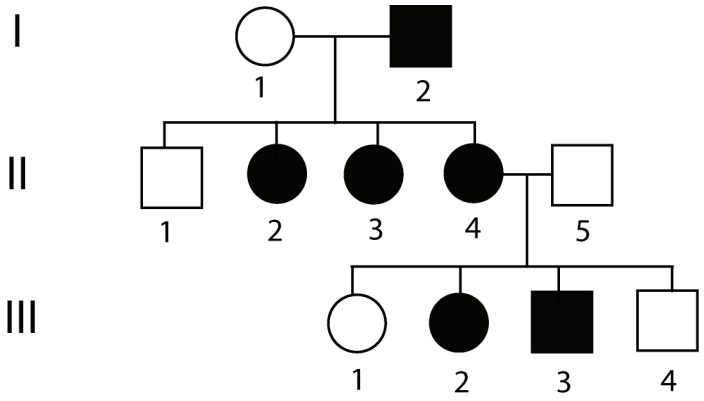
Mark allocation

- 1 mark – Huntington's disease does not take effect until reproductive maturity is reached
- 1 mark – Lethal allele can be passed on to children before condition emerges in parent(s)

Total 7 marks

Question 2

Vitamin D-resistant rickets is an inherited condition controlled by the HYP gene and is expressed even if an individual has only one copy of the allele present. The following pedigree shows a portion of a family in which some of the members have rickets. The shaded individuals on the pedigree have rickets.



2a. What is the most likely form of inheritance shown? Support your response by referring directly to the pedigree.

Solution

According to the pedigree, rickets is inherited as an X-linked dominant trait. The affected male **I-2** passes the condition to all his daughters **II-2**, **II-3** and **II-4**.

2 marks

Explanatory notes

- As a rule, for X-linked dominant traits all daughters of affected males will be affected by the condition and more females than males will show the trait. In the pedigree all the daughters of the affected male have inherited the condition and whilst the number of individuals in the pedigree is not large, there are more females than males who show the trait.

Mark allocation

- 1 mark – Rickets is an X-linked dominant trait
- 1 mark – affected male **I-2** passes the condition to all his daughters **II-2**, **II-3** and **II-4**

2b. What is the genotype of individual **II-4**?

Solution

$\mathbf{X^RX}$

1 mark

Explanatory notes

- Individual **II-4** is female as indicated by the pedigree symbol (●). Given that her partner is an unaffected male (XY) and that they produce both an unaffected daughter and son (XX; XY) and an affected daughter and son ($\mathbf{X^RX}$; $\mathbf{X^RY}$), **II-4** must be heterozygous for the trait as she can provide an $\mathbf{X^R}$ allele to one son and an X allele to the other.

2c. i. Use a Punnett square to demonstrate the outcome of a cross between individual **III-3** and an unrelated heterozygous affected female.

Solution

	(0.5) X^R	(0.5) X
(0.5) X^R	(0.25) X^RX^R	(0.25) X^RX
(0.5) Y	(0.25) X^RY	(0.25) XY

2 marks

Explanatory notes

- Punnett square must show correct gametes and proportions to gain the marks

Mark allocation

- 1 mark – correct gametes and proportions
- 1 mark – correct zygotes and proportions

2c. ii. What is the probability that the couple will produce a son who does not have Vitamin D-resistant rickets? Show your working.

Solution

Probability of a son without Vitamin D-resistant rickets is 0.25 (or 25%)

Pr(son) 0.5 × *Pr*(normal son) 0.5 = 0.25

1 mark

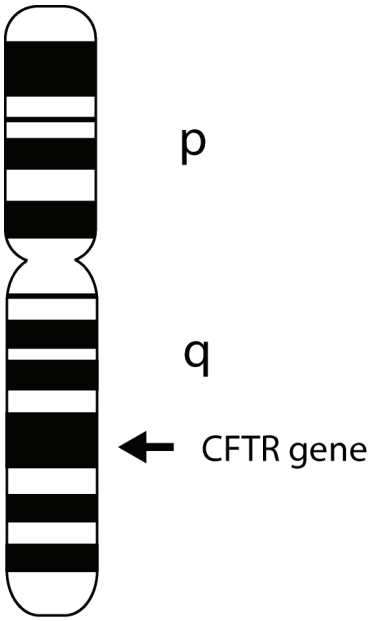
Explanatory notes

- It is necessary to consider the likelihood of **i.** conceiving a son and **ii.** conceiving a son who is normal (these are both independent events).

Total 6 marks

Question 3

Cystic fibrosis (CF) is an inherited disorder caused by a mutation of the cystic fibrosis transmembrane conductance regulator (CFTR) gene and in Australia occurs in approximately 1 in 2000 Caucasian births, with a carrier frequency of 4%. The CFTR gene is located on chromosome 7 and produces a protein (CFTR) that binds to cell membranes and acts as a channel to regulate the transport of chloride ions out of cells.



Individuals with CF demonstrate defective transport of chloride ions and secrete thick mucus which collects in and blocks airways and pancreatic ducts. Signs of CF include production of salty sweat, blockage and infection of airways and lungs, heart complications and malabsorption from the digestive tract. Individuals who are heterozygous for this mutation are not symptomatic for the condition.

3a. What form of inheritance is CF?

Solution

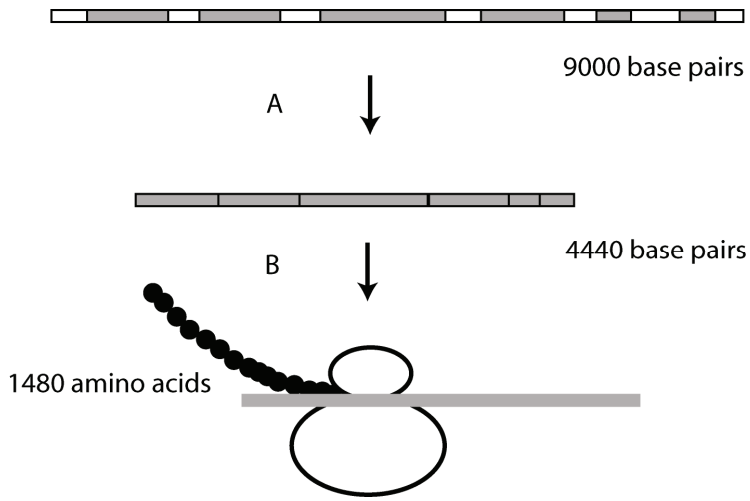
Autosomal recessive

1 mark

Explanatory notes

- The inheritance of CF is autosomal because the CFTR gene is found on chromosome 7 (not a sex chromosome) and it is recessive because heterozygotes do not show the condition. Students must justify both autosomal and recessive to achieve the mark.

The normal form of CFTR is made up of a chain of 1480 amino acids with more than 500 mutations of the CFTR gene having been described. The following diagram outlines the steps associated with the production of CFTR.



3b. i. What is the name of the process that occurs at A?

Solution

Transcription

1 mark

Explanatory notes

- mRNA nucleotide enters the nucleus to make a copy of the section of DNA that codes for CFTR.

3b. ii. What occurs between processes A and B to reduce the number of base pairs from 9000 to 4440 base pairs?

Solution

The non-coding introns have been spliced out of the pre-mRNA, leaving the coding exons which make up mRNA.

1 mark

Explanatory notes

- Students should distinguish between non-coding introns and coding exons and pre-mRNA and mRNA.

3b. iii. What is the name of the process that occurs at B?

Solution

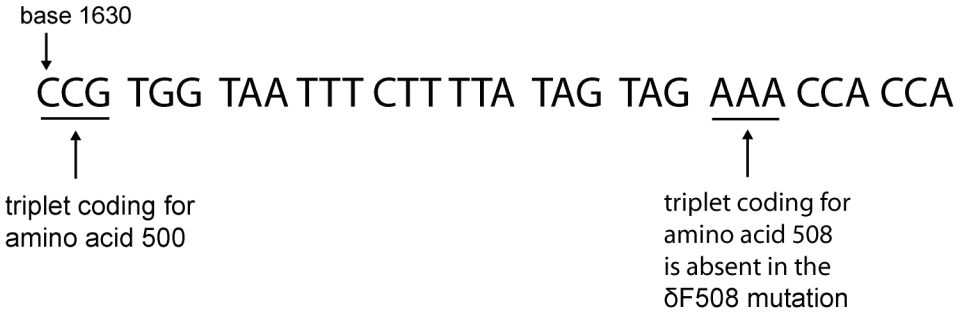
Translation

1 mark

Explanatory notes

- mRNA attaches to ribosomes and according to the coded sequence of nucleotides in the mRNA, amino acids are brought into place by tRNA molecules and a protein is assembled.

A common mutation known as the δF508 is associated with the 508th triplet in the DNA sequence of the CFTR gene and causes the absence of CFTR from its correct position in the cell membrane. Part of the DNA sequence that codes for CFTR is presented below.



3c. What type of mutation occurs at the 508th amino acid position?

Solution

Deletion mutation OR triplet deletion mutation

1 mark

Explanatory notes

- A triplet has been removed from the sequence and thus is considered to be a deletion mutation.

CF in humans has been investigated using 'knockout' mice. Experiments with knockout mice demonstrated that when copies of the normal CF gene were transferred into cells lining their airways, their symptoms of CF were reduced.

3d. What is a knockout mouse?

Solution

A mouse that has been engineered as an embryo to be missing a particular gene OR contains an inoperable or non-functional gene AND which can be used to investigate human conditions.

1 mark

Explanatory notes

- It is essential to communicate that mouse embryos are engineered so that they are missing genes. Mention of their role in the investigation of human conditions is optional.

As a result of the studies using knockout mice, clinical trials for human individuals with CF commenced. Normal CFTR genes were isolated and introduced to patients using adenoviruses (which were piped directly into lungs) and liposomes (which were inhaled as an aerosol). The human clinical trials have only shown a 25% correction of CF, with short-lived effects and in the case of the adenovirus, the death of one patient.

3e. i. What other general name is given to adenoviruses and liposomes in gene therapy?

Solution

Vector

1 mark

Explanatory notes

- A vector is an agent or virus capable of carrying passenger DNA into a cell.

3e. ii. What is a liposome and how does it deliver a normal CFTR gene to a cell?

Solution

A liposome is a hollow sphere or micelle that is capable of wrapping around fragments of DNA. When delivered to target cells, liposomes fuse with plasma membranes and deliver genes into the cell.

2 marks

Explanatory notes

- A liposome is a hollow sphere or micelle that forms spontaneously when phospholipids mix in solution. They are capable of wrapping around fragments of DNA. When delivered to target cells, liposomes fuse with the plasma membranes and deliver genes into the cell.

Mark allocation

- 1 mark – a hollow sphere or micelle
- 1 mark – liposomes fuse with plasma membranes and deliver genes into the cell

3f. Why are the effects of gene therapy for CF so temporary?

Solution

Effects of gene therapy are temporary because the normal CFTR gene is not incorporated into the DNA of chromosomes.

1 mark

Explanatory notes

- Effects of gene therapy are temporary because the normal CFTR gene is not incorporated into the DNA of chromosomes.

Total 10 marks

Question 4

Malaria is a disease which is caused by a protozoan from the genus *Plasmodium*. There are many forms of malaria, the most severe of which is caused by *Plasmodium falciparum*. Humans become infected if they are bitten by a female *Anopheles* mosquito which is carrying sporozoites in its salivary glands. The sporozoites enter the blood of the victim and infect the liver where they form merozoites. The merozoites leave the liver, enter red blood cells and reproduce by mitosis. Infected red blood cells eventually rupture and large numbers of the malarial parasite are released, along with their metabolic wastes, into the bloodstream. This results in the symptoms of chills, shaking and burning fever which are associated with malaria. The released parasites can then go on to infect more red blood cells. Drugs are available to treat malaria; however, a complete cure is difficult because the parasite is capable of remaining dormant in the liver for many years.

Travellers to countries where malaria is prevalent are advised to take anti-malarial drugs before, during and after their visit; however, this is no longer a guarantee against infection.

4a. i. Why might anti-malarial drugs no longer be successful in preventing malarial infection?

Solution

Strains of *Plasmodium* have developed a genetic resistance to the drugs.

1 mark

Explanatory notes

- Genetic variation exists within a species and whilst a particular phenotype may be selected against by the anti-malarial drugs, another may not be. Consequently the phenotype that has not been selected against will continue to reproduce and persist in the population.

4a. ii. Apart from the fact that travellers could be infected with malaria, identify another serious consequence of ineffective anti-malarial drugs.

Solution

The incidence of drug-resistant malaria in other countries will rise.

1 mark

Explanatory notes

- The incidence of drug-resistant malaria will rise because infected travellers will return to their home country and mosquitoes will spread the *Plasmodium* there.

Control of malaria has become very difficult and much research has been directed towards finding a successful strategy that will eradicate the disease. Recently scientific trials have resulted in the production of transgenic mosquitoes which cannot pass on malaria.

4b. What is the difference between a genetically modified organism and a transgenic organism?

Solution

A genetically modified organism is any organism whose genetic make-up has been artificially changed (and includes transgenic organisms) whereas a transgenic organism is one that carries in its genome a gene or genes that have been artificially introduced *from another species*.

2 marks

Mark allocation

- 1 mark – GMO is any organism whose genetic makeup has been artificially changed
- 1 mark – TGO carries a gene or genes artificially introduced from another species

4c. What are two techniques that might be used to develop transgenic organisms?

Solution

Any **two** of the following:

- Micro-injection of DNA of the gene into the host cell (somatic or egg cell)
- Transfer using a virus (adenovirus or retrovirus)
- Ballistics ('gene gun')
- Electric pulse

1 mark

Explanatory notes

- Students do not need to describe these techniques but they should be aware of their names. If only one technique is given, 0 marks are awarded.

In a study using transgenic mosquitoes, researchers combined equal numbers of transgenic and wild-type mosquitoes which were fed on malaria-infected mice. The mice were infected with *Plasmodium* – the parasite that causes malaria in humans. After nine generations, the number of mosquitoes in the two populations was compared.

	Generation 1	Generation 9
Transgenic mosquitoes	0.5	0.7
Wild-type mosquitoes	0.5	0.3

4d. i. What is being described by the numerical value attributed to the transgenic and the wild-type mosquitoes at Generation 9?

Solution

The fitness value OR the frequency of each phenotype of mosquito population in the generation

1 mark

Explanatory notes

- If a phenotype makes a greater contribution to the gene pool in the next (and subsequent) generations, it is said to have a higher fitness value or to be at a selective advantage. The phenotype that makes a lesser contribution is said to be less fit or to be selected against.

4d. ii. Identify one factor that might have contributed to the greater proportion of the transgenic mosquitoes.

Solution

They might have had a higher survival rate OR they might have laid more eggs.

1 mark

Total 7 marks

Question 5

Neofelis diardi, also known as the Bornean clouded leopard, lives on the South-east Asian islands of Borneo and Sumatra and has been identified as a new species of leopard. Clouded leopards were first documented in 1821 and *Neofelis nebulosa* is the species found on the South-east Asian mainland. Until recently the mainland and island animal were thought to have been the same species, however DNA tests highlighting around 40 nucleotide differences between the two species (lions and leopards have 56 nucleotide differences) suggest the two species separated 1.4 million years ago. Results from the genetic study are also supported by separate research based mainly on fur patterns and colouration of skins. The new clouded leopard species is generally darker than the mainland species, has small cloud markings, many distinct spots within the cloud markings, greyer fur and a double dorsal stripe. Clouded leopards from the mainland have large clouds on their skin with fewer, often faint, spots within the cloud markings and they are lighter in colour, with a tendency toward tawny-coloured fur and a partial double dorsal stripe.

5a. How could the ancestors of the Bornean leopard have arrived in Borneo and Sumatra from the South-east Asian mainland?

Solution

They crossed a now submerged land bridge OR by rafting.

1 mark

Explanatory notes

- The only realistic possibilities for this response would be as stated above. Whilst leopards are capable of swimming, they are not known for swimming long distances. It should be remembered that the leopards diverged 1.4 million years ago and therefore any human involvement (boats) in their migration is impossible.

5b. What kind of speciation has occurred in the clouded leopard?

Solution

Allopatric speciation

1 mark

Explanatory notes

- Geographical fragmentation has occurred between the South-east Asian mainland and the islands of Borneo and Sumatra, thus speciation is due to allopatric speciation.

5c. Explain how the differences between the two populations of clouded leopard have come about.

Solution

A smaller group *of individuals splits from a parent population* and becomes *geographically isolated*. *Over many generations* the geographically isolated population is subjected to *different selection pressures* due to different environmental conditions and genetic drift. *Over time, the isolated population changes* so much that even if they were to come together again, they would *not be able to reproduce successfully*.

2 marks

Explanatory notes

- It is useful to remember the process of speciation as a series of steps. This will mean that students will remember the sequence if required.
  Step 1: Migration
  Step 2: Geographical isolation
  Step 3: Different selection pressures
  Step 4: Reproductive isolation

Mark allocation

- 1 mark – migration leading to geographical isolation
- 1 mark – different selection pressures leading to reproductive isolation, hence speciation

5d. What form of evolution has occurred in the speciation of the Bornean leopard?

Solution

Divergent evolution

1 mark

Explanatory notes

- The diversification of an ancestral group into two or more species in different habitats is referred to as divergent evolution. This is what has occurred in the case of the clouded leopard on mainland South-east Asia and the islands of Borneo and Sumatra.

Total 5 marks

Question 6

Proteins are biological molecules, the basic unit of which is an amino acid. The production of all proteins is genetically controlled and any difference in the sequence of amino acids is indicative of a change in DNA sequence.

The following table shows the number of differences that exist in the amino acid sequence of the beta chain of haemoglobin in humans and other vertebrates. Haemoglobin is the protein in red blood cells that is responsible for oxygen transport. In adults, the haemoglobin molecule is comprised of four polypeptide chains, 2 alpha chains consisting of 141 amino acids and 2 beta chains consisting of 146 amino acids.

Vertebrate	No. of amino acids different from humans
gorilla	1
gibbon	3
rhesus monkey	8
mouse	27
chicken	45
frog	67

6a. Which vertebrate is most closely related to humans?

Solution

The gorilla as there is only one difference between humans and gorillas in terms of the amino acid sequence of the beta chain of haemoglobin.

1 mark

Explanatory notes

- There is only one difference between humans and gorillas in terms of the amino acid sequence of the beta chain of haemoglobin; there are greater differences in the remaining vertebrates.

6b. Why can the comparison of amino acid differences in proteins be used to determine how closely different vertebrates are related?

Solution

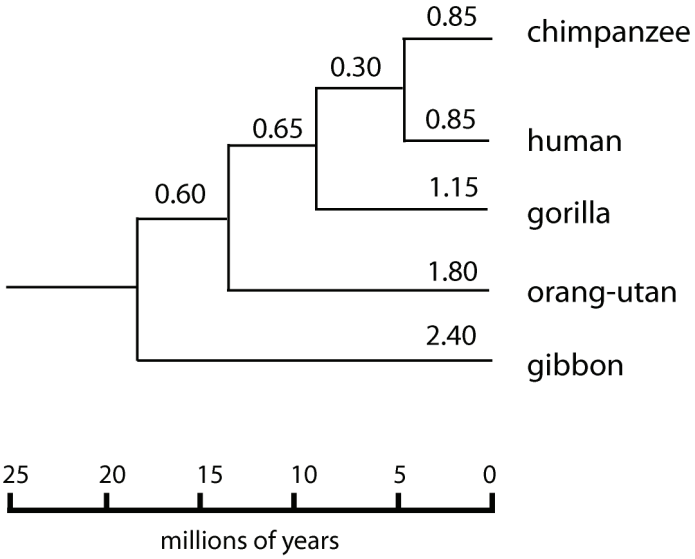
Species that are more closely related are expected to have fewer differences in their amino acid sequences for corresponding proteins.

1 mark

Explanatory notes

- Species that are more closely related are expected to have fewer differences in the amino acid sequences of corresponding proteins than species that are more distantly related. This is because the longer the time that has elapsed since the two species diverged from their last common ancestor, the longer the time that has passed in which changes could occur in the protein of both species.

The following diagram is an evolutionary tree showing the inferred relationships between groups of primates based on DNA hybridisation evidence.



6c. How can DNA hybridisation provide a measure of genetic relatedness between species?

Solution

The similarity of DNA can be determined by measuring how closely single strands of DNA from each species combine. The more similar the DNA, the higher the degree of relatedness and the more difficult it is to separate the hybridised DNA.

2 marks

Explanatory notes

- DNA–DNA hybridisation is a technique which is used to compare the similarity of genomes of different species and by doing so, infer their degree of evolutionary relationship.

Mark allocation

- 1 mark – by measuring how closely single strands of DNA from each species combine
- 1 mark – the more similar the DNA, the higher the degree of relatedness and the more difficult it is to separate the hybridised DNA

From the diagram it is possible to infer that the gibbon line diverged around 20 million years ago. After completion of the DNA–DNA hybridisation and in order to be more certain about time scales, it is essential to calibrate the evolutionary tree against some other measure.

6d. In order to identify a time scale for this evolutionary tree, what other form of evidence would be used for calibration?

Solution

Fossil evidence

1 mark

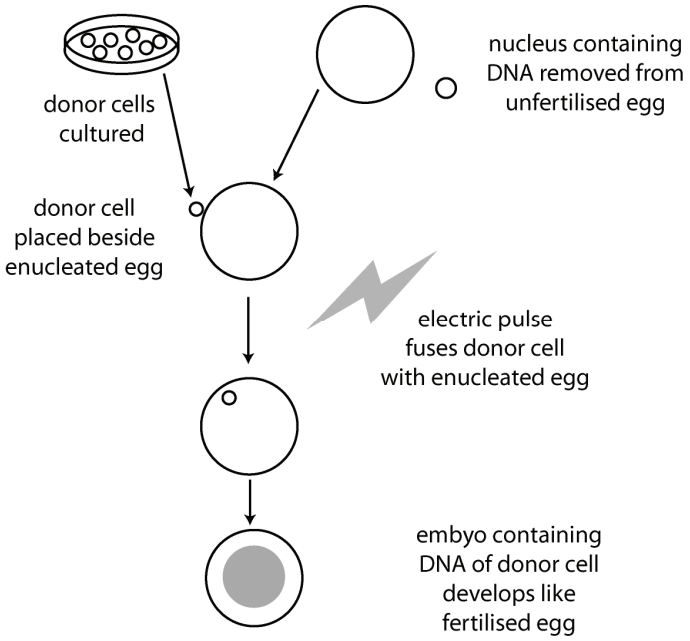
Explanatory notes

- DNA–DNA hybridisation provides a measure of the similarity of the genetic material of two species and provides an estimate of their genetic distance, however it does not provide an indication of time scale. This can be identified by comparing the hybridisation results against the fossil record.

Total 5 marks

Question 7

Cloning is the process of producing genetically identical organisms from one parent. There are two known techniques for cloning an organism. The following diagram outlines the technique used to produce the Afghan hound 'Snuppy' (named after the **S**eoul **N**ational **U**niversity p**uppy**), the first dog to be artificially cloned from the ear cell of a 3-year-old Afghan hound.



7a. i. What is the name of the reproductive cloning technique shown in the diagram?

Solution

Nuclear transfer

1 mark

Explanatory notes

- The two techniques used for cloning are nuclear transfer and embryo splitting. From the diagram it is clear that a donor cell containing a nucleus has been transferred into an enucleated cell, thus the technique can only be nuclear transfer.

7a. ii. Explain how the technique you have identified in **7ai.** differs from therapeutic cloning.

Solution

The purpose of reproductive cloning is to produce a new organism whereas the purpose of therapeutic cloning is to produce stem cells.

1 mark

Explanatory notes

- Reproductive cloning seeks to produce a new organism via the techniques of nuclear transfer or embryo splitting. Therapeutic cloning however, uses the technique of nuclear transfer to create an embryo from which stem cells can be harvested for use in treating spinal cord or brain injuries, stroke or degenerative diseases, for example.

7a. iii. How might the application of therapeutic cloning affect the evolutionary process?

Solution

Normal evolutionary pressures of natural selection will no longer be acting on organisms with particular disorders.

1 mark

Explanatory notes

- Therapeutic cloning has the potential to alleviate or repair damage which results from disease or disorder. It treats the condition but is only a somatic remedy. If the condition is heritable, therapeutic cloning will not treat germ line cells. The organism free from symptoms may survive to reproductive age and pass their faulty alleles into the next generation. Ordinarily, due to natural selection, the organism would have been at a selective disadvantage, but with therapeutic cloning it will not.

In order to produce Snuppy, 123 embryos were implanted into surrogate females, only three embryos survived. Of these embryos one died just prior to birth, another died soon after birth and the third was the only survivor – Snuppy. Clearly the success of live births from cloning is not high.

7b. What is another disadvantage associated with this cloning technique?

Solution

Severe deformities OR disease associated with premature ageing

1 mark

It has been hypothesised that animals cloned from adult somatic cells will age more rapidly than those which have been cloned from younger somatic cells.

7c. Why might animals cloned from adult somatic cells age more rapidly?

Solution

Their telomeres become shorter after each mitotic division; this is a loss of DNA and is linked with ageing.

1 mark

Explanatory notes

- Each time mammalian cells divide they lose DNA base pairs from the telomeres (ends of the chromosomes). This is thought to be associated with ageing.

Total 5 marks

Question 8

Over time, human populations have been affected by change which has resulted in the variations observable between different indigenous populations. Biological evolution is one type of change that has occurred in the human population and has affected inherited phenotypes.

8a. What is the name given to the process through which biological evolution occurs and what is its outcome?

Solution

Natural selection is a process which leads to differential survival and reproduction.

1 mark

Explanatory notes

- The process through which human evolution occurs is known as natural selection and it produces differential survival and reproduction under particular environmental conditions.

In humans, the ability to digest lactose, a sugar found in milk, is due to the presence of an enzyme. Individuals who can digest lactose are considered to be 'lactose tolerant' while those who cannot are 'lactose intolerant'. Mammals are usually lactose tolerant, with humans showing tolerance up to the age of four years. Beyond this age however, lactose intolerance becomes increasingly common. Lactose intolerance results in abdominal cramps, bloating and diarrhoea and occurs soon after consuming fresh milk or fresh milk products.

8b. Is the human population monomorphic or polymorphic in terms of its ability to digest lactose? Explain your answer.

Solution

The human population is polymorphic in terms of its ability to digest lactose because there are two discrete variations that exist – lactose intolerant and lactose tolerant.

1 mark

Explanatory notes

- A population that is polymorphic for a trait will demonstrate two or more discrete variations of that trait, whereas a population that is monomorphic will only exhibit a single expression of that trait.

8c. Humans have not always been lactose tolerant. What is the most likely cause of lactose tolerance?

Solution

Lactose tolerance is most likely due to a gene mutation.

1 mark

Domestication of wild cattle occurred as the first human communities established agricultural practices.

8d. At this time, would one of the phenotypes have been at a selective advantage? Explain your answer.

Solution

The lactose-tolerant phenotype would have been at a selective advantage because individuals with this phenotype would have been able to incorporate the food as part of their nutrient intake.

2 marks

Explanatory notes

- The lactose-tolerant phenotype would have been at a selective advantage because individuals with this phenotype would have been able to incorporate the food as part of their nutrient intake. This would increase the likelihood of surviving to successfully reproduce.

Mark allocation

- 1 mark – lactose-tolerant phenotype would have been at a selective advantage
- 1 mark – individuals with this phenotype would have been able to incorporate the food as part of their nutrient intake

Total 5 marks

END OF SOLUTIONS BOOK