INSIGHT

YEAR 12 *Trial Exam Paper*

2012
BIOLOGY
Written examination 2

Solutions book

This book presents:

- correct solutions
- explanatory notes
- mark allocations

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SECTION A – Multiple-choice questions

Question 1

A recessively inherited condition is expressed only in individuals with type O blood. A man with type A blood has a child with a woman with type B blood. Their first child is born with the condition even though it is not expressed in either of the parents. Both the parents are heterozygous for the gene that causes the disease. What is the probability that their second child will have the condition?

A. $\frac{1}{4}$

B. $\frac{1}{16}$

C. $\frac{1}{3}$

D. $\frac{3}{4}$

Answer is B

Explanatory notes

- A is incorrect – of the 16 possible outcomes from this cross, there is only one chance that their child will have the condition, NOT $\frac{1}{4}$.
- B is correct – of the 16 possible outcomes from this cross, there is only one chance that their child will have the condition.
- C is incorrect – of the 16 possible outcomes from this cross, there is only one chance that their child will have the condition, NOT $\frac{1}{3}$.
- D is incorrect – of the 16 possible outcomes from this cross, there is only one chance that their child will have the condition, NOT $\frac{3}{4}$.

Question 2

In sesame plants, pod type and leaf type are inherited independently. The one-pod condition (*P*) is dominant to the three-pod condition (*p*), and normal leaf (*L*) is dominant to wrinkled (*l*). A cross between two plants produces the following offspring:

273 one-pod, normal leaf	289 one-pod, wrinkled leaf	67 three-pod, normal leaf	56 three-pod, wrinkled leaf

The cross that would produce these results is

A. *PpLl* × *PpLl*.

B. *ppLl* × *ppLl*.

C. *PPll* × *ppLL*.

D. ***PpLl* × *Ppll*.**

Answer is D

Explanatory notes

- A is incorrect – this cross would produce results in a 3:2:2:1 ratio.
- B is incorrect – this cross would produce results in a 3:1 ratio.
- C is incorrect – all the offspring produced from this cross would be identical for these two traits.
- D is correct – variation does not occur as a result of mitosis.

Question 3

In humans, the genes of the major histocompatibility complex (MHC) code for human leukocyte antigens (HLA) as well as other proteins. HLA proteins are present on the surface of most body cells. The MHC consists of more than 70 genes, classified into class I, II, and III MHC. There may be as many as one hundred different alleles at a single locus. An HLA gene could be described as

A. **polymorphic.**

B. polygenic.

C. monomorphic.

D. monogenic.

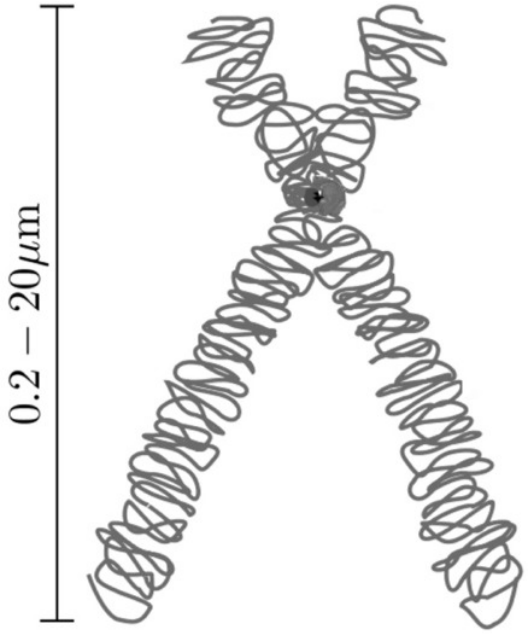
Answer is A

Explanatory notes

- A is correct – a polymorphism is associated with several variants of a particular trait.
- B is incorrect – polygenic describes traits that are controlled by the action of two or more polygenes.
- C is incorrect – monomorphic refers to a population in which all individuals are identical with respect to a specific trait.
- D is incorrect – monogenic describes traits that are under the control of single genes.

The following information relates to Questions 4 and 5.

The image shows a diagram of a human chromosome.



Chromosome: Image by Magnus Manske

Question 4

The chromosome shown is duplicated and as a result has

A. one arm.

B. two arms.

C. three arms.

D. **four arms.**

Answer is D

Explanatory notes

- A is incorrect – there are four arms, 2 p-arms and 2 q-arms, NOT one.
- B is incorrect – there are four arms, 2 p-arms and 2 q-arms, NOT two.
- C is incorrect – there are four arms, 2 p-arms and 2 q-arms, NOT three.
- D is correct – there are four arms, 2 p-arms and 2 q-arms.

Question 5

When a cell is not dividing, the chromosomes are present as

A. microtubules.

B. tubulin fibres.

C. **chromatin fibres.**

D. kinetochores.

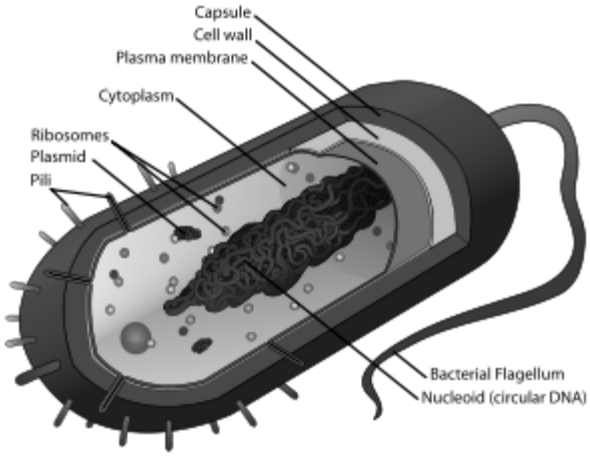
Answer is C

Explanatory notes

- A is incorrect – microtubules are hollow rods made of tubulin found in the cytoplasm of eukaryotes.
- B is incorrect – tubulin fibres are globular proteins that make up microtubules.
- C is correct – chromatin is the complex of DNA and proteins that make up a eukaryote chromosome.
- D is incorrect – a kinetochore is a structure of proteins attached to the centromere that connects the sister chromatids to the mitotic spindle.

Question 6

The diagram shows a prokaryote.



Which of the following best describes the genetic material of the prokaryote?

	Plasmid	**Chromosome**
A.	single-stranded DNA	single-stranded DNA
B.	**double-stranded DNA**	**double-stranded DNA**
C.	single-stranded RNA	single-stranded RNA
D.	double-stranded RNA	double-stranded RNA

Answer is B

Explanatory notes

- A is incorrect – the DNA of all prokaryotes is double stranded, NOT single stranded.
- B is correct – the DNA in plasmids and chromosomes of all prokaryotes is double stranded.
- C is incorrect – plasmids of prokaryotes are made of DNA, NOT RNA and the DNA is double stranded, NOT single stranded.
- D is incorrect – plasmids of prokaryotes are made of DNA, NOT RNA.

Question 7

Hydrangea: Image by Joachim Alves Gaspar

The hydrangea (*H. macrophylla*) is a plant characterised by a large flower-heads. The flowers can be blue, red, pink, light purple or dark purple depending on the pH of the soil. Anthocyanidins pigments found commonly in plants, are responsible for colouring hydrangeas red in acid soil and blue in alkaline soil. The variation in flower colour of hydrangeas is under

A. genetic control.

B. environmental control.

C. **genetic and environmental control.**

D. genetic, environmental and GM control.

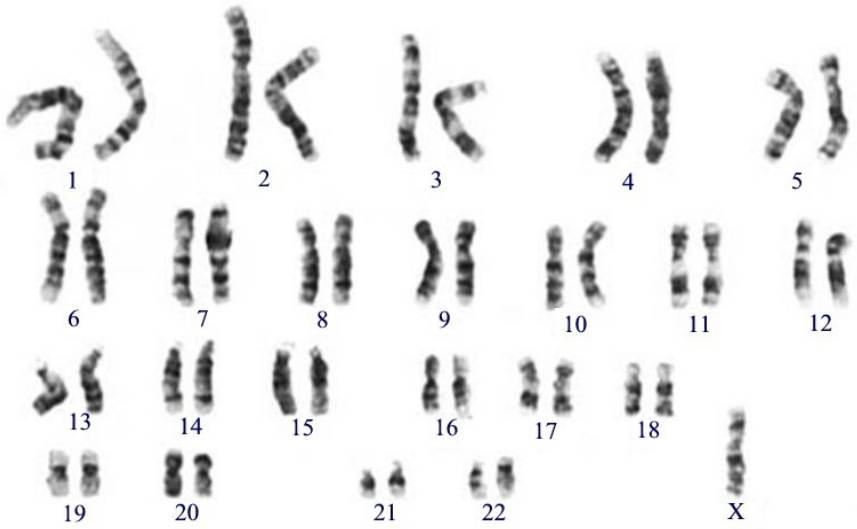
Answer is C

Explanatory notes

- A is incorrect – variation in flower colour is due to genetic and environmental control, NOT just genetic control.
- B is incorrect – variation in flower colour is due to genetic and environmental control, NOT just environmental control.
- C is correct – variation in flower colour is due to genetic and environmental control.
- D is incorrect – variation in flower colour is due to genetic and environmental control, NOT GM control.

The following information relates to Questions 8 and 9.

Question 8



The condition shown in the karyotype occurs as a result of

A. **monosomy.**

B. trisomy.

C. translocation.

D. inversion.

Answer is A

Explanatory notes

- A is correct – there is a complete absence of one sex chromosome, making this a monosomy.
- B is incorrect – trisomy involves the inclusion of an extra chromosome in a karyotype.
- C is incorrect – translocation is due to the rearrangement of parts between non-homologous chromosomes.
- D is incorrect – an inversion occurs when a single chromosome is reversed end to end.

Question 9

From the karyotype it is possible to describe the sex of the baby as

A. male.

B. **female.**

C. intersex.

D. undetermined.

Answer is B

Explanatory notes

- A is incorrect – the absence of a Y-chromosome rules out male.
- B is correct – the presence of an X-chromosome confers female.
- C is incorrect – an intersex karyotype would show an X-chromosome and a Y-chromosome, but it is not possible to determine whether or not the baby is intersex on the basis of a karyotype alone.
- D is incorrect – the presence of an X-chromosome means the baby has a sex, which is female.

Question 10

The amino acid leucine can be produced by any of the following triplets:

AAT AAC GAA GAG GAT GAC

A tRNA molecule that codes for the amino acid leucine is

A. AAT.

B. CTC.

C. GCU.

D. **GAU.**

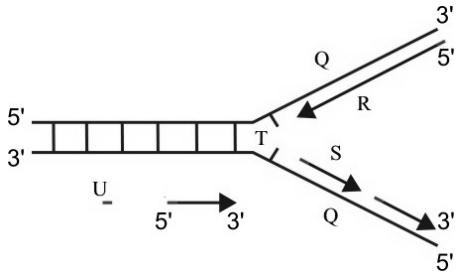
Answer is D

Explanatory notes

- A is incorrect – there will be no thymine (T) in tRNA.
- B is incorrect – there will be no thymine (T) in tRNA.
- C is incorrect – the DNA codon that corresponds with GCU is GCT. This does not code for leucine.
- D is correct – the DNA codon that corresponds with GAU is GAT.

Question 11

The diagram shows a DNA replication fork.



The correct labelling for the diagram is

	Q	R	S	T	U
A.	DNA template	lagging strand	leading strand	replication fork	Okazaki fragment
B.	DNA template	leading strand	lagging strand	Okazaki fragment	primer
C.	**DNA template**	**leading strand**	**lagging strand**	**replication fork**	**primer**
D.	DNA template	lagging strand	leading strand	primer	Okazaki fragment

Answer is C

Explanatory notes

- A is incorrect – R is a leading strand, not a lagging strand.
- B is incorrect – T is a primer and U is an Okazaki fragment, not the other way around.
- C is correct – all labels are correct.
- D is incorrect – R is a leading strand and S is a lagging strand, not the other way around, T is a replication fork, not a primer and U is a primer, not an Okazaki fragment.

The following information relates to Questions 12 and 13.

The mummichog fish (*Fundulus heteroclitus*) is a particularly hardy fish found along the eastern seaboard of the United States and Atlantic Canada. The mummichog fish requires a liver enzyme, lactate dehydrogenase (LDH) to maintain a high metabolic rate. A high metabolic rate enables the fish to maintain a high body temperature so they can survive and reproduce in cold waters. The diagram shows that in mummichog fish, the frequency of the allele for LDH decreases in fish sampled from Maine to Georgia. The table shows the relationship between genotype and phenotype in terms of the level of LDH in the liver.

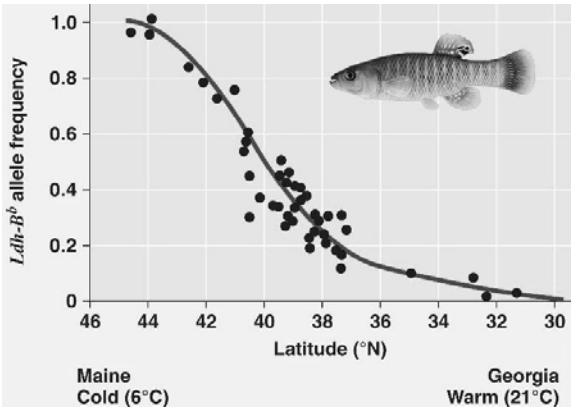


Image from Campbell, Reece, Meyers et al. Biology 8th edn

Genotype	Phenotype (level of LDH enzyme in the liver)
BB	4 units/g liver
Bb	2 units/g liver
bb	1 unit/g liver

Question 12

The range of frequencies for LDH in mummichog fish is an example of

A. **geographical variation.**

B. developmental variation.

C. behavioural variation.

D. structural variation.

Answer is A

Explanatory notes

- A is correct – this is an example of geographical variation, specifically, a cline.
- B is incorrect – this is not an example of developmental variation.
- C is incorrect – this is not an example of behavioural variation.
- D is incorrect – this is not an example of structural variation.

Question 13

By chance in one of the liver cells of a fish with the genotype *bb* there is a mutation that results in the change of a *b* allele for LDH into a *B* allele. This is unlikely to assist the fitness of the fish because

A. there will be no change in the amount of LDH produced in the liver.

B. the mummichog fish is already producing the maximum level of LDH.

C. the change in genotype will cause the rate of metabolism to decrease further.

D. the mutation has taken place in a somatic cell of the liver and will not raise the LDH sufficiently to effect a change.

Answer is D

Explanatory notes

- To improve the fitness of the fish, the mutation would need to take place in a gamete so that the trait can be passed on to subsequent generations.
- A is incorrect – there will be a miniscule change in the amount of LDH but not enough to improve the fitness of the fish.
- B is incorrect – the mummichog fish is not producing the maximum level of LDH.
- C is incorrect – the change in genotype will result in a miniscule change in the amount of LDH but not enough to improve the fitness of the fish.
- D is correct – the mutation has taken place in a somatic cell of the liver and will not raise the LDH sufficiently to improve the fitness of the fish.

Question 14

Snakes are capable of an impressive form of cranial kinesis whereby they are able to move the bones in their upper jaw to enable them to swallow prey much larger than their own head. This is an example of an adaptive evolution. The only evolutionary mechanism that consistently leads to adaptive evolution is

A. gene flow.

B. **natural selection.**

C. genetic drift.

D. founder effect.

Answer is B

Explanatory notes

- A is incorrect – gene flow can cause the frequency of advantageous alleles to increase or decrease. It does not consistently lead to adaptive evolution.
- B is correct – natural selection only ensures the survival of the genetically most fit and therefore consistently leads to adaptive evolution.
- C is incorrect – genetic drift can cause the frequency of advantageous alleles to increase or decrease. It does not consistently lead to adaptive evolution.
- D is incorrect – founder effect is a form of genetic drift; genetic drift can cause the frequency of advantageous alleles to increase or decrease. It does not consistently lead to adaptive evolution.

Question 15

In eukaryote organisms the greatest source of genetic variation is due to

A. polymorphism.

B. masking of recessive alleles.

C. **recombination.**

D. mutations.

Answer is C

Explanatory notes

- A is incorrect – genetic variation leads to polymorphism in a population, not vice versa.
- B is incorrect – genetic variation occurs regardless of whether recessive alleles are masked (or not).
- C is correct – recombination of chromosomes during meiosis is the greatest source of genetic variation.
- D is incorrect – mutations do contribute to genetic variation, however, they are either rare, have no effect or are deleterious, so they do not often persist within a population and genetic variation cannot therefore be attributed to them.

Question 16

Humans (*Homo sapiens*) and dogs (*Canis lupus familiaris*) have been living together for over 10,000 years. The images below show some of the dogs that exist today.

Mexican Wolf
Image by Trisha Shears

Chihuahua mix and Great Dane
Image by: Rogrido de Almeida

Shar pei
Image by Ellen Levy Finch

This variation in breeds is due to

A. **artificial selection.**

B. natural selection.

C. disruptive selection.

D. stabilising selection.

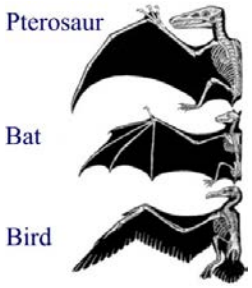
Answer is A

Explanatory notes

- A is correct – artificial selection is intentional breeding in which humans favour certain traits, or combinations of traits.
- B is incorrect – natural selection is the process by which agents of selection act differentially on specific phenotypes in members of a population. There is no human involvement.
- C is incorrect – disruptive selection is a form of natural selection in which individuals at both extremes of a phenotypic range survive or reproduce more successfully than individuals with an intermediate phenotype.
- D is incorrect – stabilising selection is a form of natural selection in which intermediate phenotypes survive or reproduce more successfully than individuals at the extremes of a phenotypic range.

Question 17

Consider the following diagram.



The wings of pterosaurs, bats and birds are

A. analogous as forearms.

B. homogenous as forearms.

C. **analogous as wings.**

D. homologous as wings.

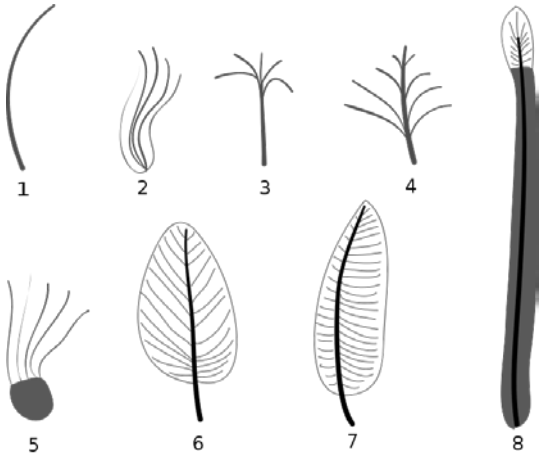
Answer is C

Explanatory notes

- A is incorrect – the wings of pterosaurs, bats and birds are analogous as wings but homologous as forearms.
- B is incorrect – the wings of pterosaurs, bats and birds are homologous as forearms, NOT homogenous.
- C is correct - the wings of pterosaurs, bats and birds are analogous as wings.
- D is incorrect – the wings of pterosaurs, bats and birds are homologous as forearms, NOT wings.

The following information relates to Questions 18 and 19.

When feathers first appeared in the fossil record, it is clear that they were not used for airborne travel. The first feathers were hairlike, hollow filaments. More mutations occurred, leading to branching of filaments which then joined together. The following diagram shows the stages of evolution in feathers.



Feather stages: Image by Matt Martyniuk

1 Single filament
2 Multiple filaments joined at their base
3 Multiple filaments joined at their base to a central filament
4 Multiple filaments along the length of a central filament
5 Multiple filaments arising from the edge of a membranous structure
6 Pennaceous feather with vane of barbs and barbules and central rachis
7 Pennaceous feather with an asymmetrical rachis
8 Undifferentiated vane with central rachis

Question 18

It would be reasonable to expect that in the fossil record there will be

A. no dinosaurs with feather-like coverings.

B. **dinosaurs with feather-like coverings of many kinds.**

C. dinosaurs with one kind of feather-like covering.

D. only one type of dinosaur with one kind of feather-like covering.

Answer is B

Explanatory notes

- A is incorrect – feathers appeared in the fossil record during the Cretaceous period (dinosaurs).
- B is correct – there were dinosaurs with many different types of feather-like coverings in the fossil record.
- C is incorrect – there were dinosaurs with many different types of feather-like coverings in the fossil record, NOT just one kind.
- D is incorrect – there were many dinosaurs with many different types of feather-like coverings in the fossil record, NOT just one kind.

Question 19

Which of the following statements about feathers is NOT correct?

Feathers

A. play a role in mate selection in birds.

B. **evolved to enable dinosaurs to fly.**

C. are associated with thermoregulation.

D. assist in camouflage.

Answer is B

Explanatory notes

- A is incorrect – the stot feathers do play a role in mate selection.
- B is correct – feathers evolved by chance: NOT for a purpose.
- C is incorrect – this statement is true: feathers are associated with thermoregulation.
- D is incorrect – this statement is true: feathers do assist in camouflage.

Question 20

The order of fossils in rock strata indicates the sequence in which the fossils were laid down. It does not however provide information about their actual ages. Which of the following techniques could be used to determine the actual age of a fossil?

A. stratigraphy

B. biostratigraphy

C. lithology

D. **radiometry**

Answer is D

Explanatory notes

- A is incorrect – stratigraphy is the branch of geology which studies rock layers and layering; it produces relative data, not absolute.
- B is incorrect – biostratigraphy is the branch of stratigraphy which focuses on correlating and assigning relative ages of rock strata by using the fossil assemblages contained within them; it produces relative data, not absolute.
- C is incorrect – lithology is a description of the physical characteristics of rock such as colour, texture, grain size or composition; it produces relative data, not absolute.
- D is correct – radiometry is a set of techniques for measuring electromagnetic radiation and provides absolute data about the age of rocks.

The following information relates to Questions 21 and 22.

In 2011, scientists published a paper claiming that when humans eat Asian rice (*Oryza sativa*), a microRNA (miRNA) highly expressed in that rice is secreted into vesicles during digestion and then taken up by liver cells, where the miRNAs then have a physiological effect on human low-density lipoprotein (LDL) levels. All eukaryote organisms produce miRNAs, tiny non-coding sequences of RNA (only 22 nucleotides long). They enable a cell to control how much of a specific protein it produces, allowing rapid and flexible, real-time responses to changing conditions. Subsequent research has shown that the miRNA from the rice could inhibit uptake of LDLs, which can lead to atherosclerosis.

Without any scientific evidence, a writer linked miRNAs with genetically modified organisms (GMOs), in an attempt to alert people to the danger that manipulating plant DNA will lead to an increase in human diseases such as cancer, Alzheimer's disease and diabetes.

GMOs were not used in the Asian rice study.

Question 21

A GMO is an organism

A. **whose genome has been artificially changed.**

B. whose genome has been altered by recombination during meiosis.

C. which has been cloned.

D. which has been exposed to mutagens.

Answer is A

Explanatory notes

- A is correct – a GMO is an organism whose genome has been artificially changed and also includes TGOs.
- B is incorrect – recombination through meiosis occurs naturally and will therefore not produce a GMO.
- C is incorrect – a clone is an organism which is genetically identical to another, it is not necessarily a GMO.
- D is incorrect – exposure to mutagens does not produce a GMO.

Question 22

Theoretically miRNAs from Asian rice are not harmful to humans because

A. they are small molecules.

B. they are comprised of nucleic acid.

C. **miRNAs are present in everything we ingest.**

D. they are enclosed in vesicles which do not break open.

Answer is C

Explanatory notes

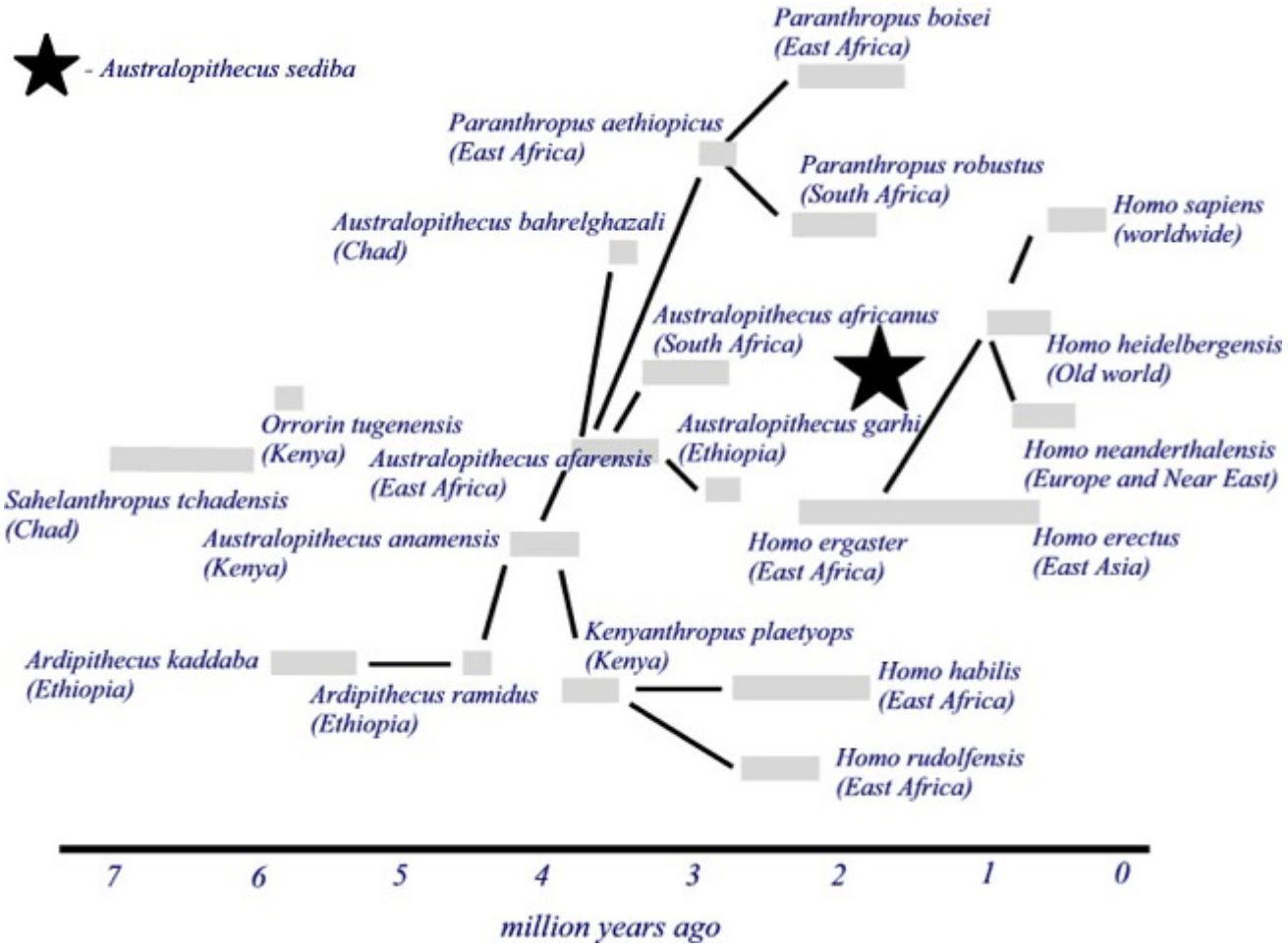
- A is incorrect – small molecules (viruses, prions) can be harmful to humans.
- B is incorrect – nucleic acid can be harmful to humans (viral DNA/RNA).
- C is correct – humans have constant regular intake of material from eukaryotes.
- D is incorrect – vesicles can open to release their content; this is how they enter a cell.

The following information relates to Questions 23, 24 and 25.

In September 2011, researchers revealed new details about the brain, pelvis, hands and feet of *Australopithecus sediba* (*A. sediba*), a primitive hominin that existed around the same time early *Homo* species first began to appear on Earth. The fossils of a juvenile male (MH-1) and an adult female (MH-2) *A. sediba* were found together in the Malapa caves in South Africa in 2010.

The fossil record for early *Homo* is very ambiguous. Many fossils are attributed to several species and their dating can be inaccurate. *A. sediba* has a number of derived characteristics, which it shares with the genus *Homo*. *A. sediba* has many more traits in common than any other early *Homo* (*H. rudolfensis*, *H. habilis*) with *H. erectus*.

The diagram shows an interpretation of the evolutionary relationship of the genus *Homo*.



Question 23

The diagram suggests that *A. sediba* is likely to be

A. a descendant of *H. rudolfensis*.

B. an ancestor of *A. afarensis*.

C. a descendant of the first species of the *Homo* genus, *H. erectus*.

D. an ancestor of the first species of the *Homo* genus, *H. erectus*.

Answer is D

Explanatory notes

- A is incorrect – *A. sediba* has traits of an Australopithecine and *H. rudolfensis* does not have any known descendants.
- B is incorrect – *A. sediba* has been dated as younger than *A. afarensis* and can only be considered a descendant.
- C is incorrect – being an Australopithecine, *A. sediba* cannot be a descendant of *H. erectus*.
- D is correct – being an Australopithecine, *A. sediba* can only be an ancestor of *H. erectus*.

Question 24

The wrist and hand of MH-2 were only missing a few bones, making them the most complete hand fossils for an early hominin on record. The hand showed a strong flexor apparatus (which hints at tree-climbing) and also had a long thumb and short fingers (a sign of precision gripping). Considering the long thumb and short fingers of the hand it is highly likely that *A. sediba*

A. was highly adept at tree climbing.

B. **engaged in tool making.**

C. was an agile quadruped.

D. was skilled at cave painting.

Answer is B

Explanatory notes

- A is incorrect – tree climbers have long fingers for grasping. *A. sediba* has short fingers and while it may have had strong muscles for climbing trees, it would not have been highly adept.
- B is correct – precision grip enables manipulation of objects and use of tools, thus *A. sediba* would have been a tool maker.
- C is incorrect – Australopithecines were bipedal, thus *A. sediba* would NOT have been a quadruped.
- D is incorrect – the earliest cave painting has been dated at around 35,000 years ago, therefore *A. sediba* could not have been skilled at cave painting.

Question 25

What radiometric dating method would have been used to date the age of *A. sediba*?

A. **uranium–lead dating**

B. carbon-14 dating

C. electron-spin resonance

D. potassium–argon dating

Answer is A

Explanatory notes

- A is correct – uranium–lead dating is used to date igneous rocks in the range from 1000 to 1,000,000 years old.
- B is incorrect – carbon-14 dating is used to date organic remains from the present to 60,000 years ago. *A. sediba* is much older.
- C is incorrect – electron-spin resonance is used to date organic material from 50,000 to 500,000 years old. *A. sediba* is much older.
- D is incorrect – potassium–argon dating is used to date igneous rocks containing K-bearing minerals. *A. sediba* was not found in this kind of rock.

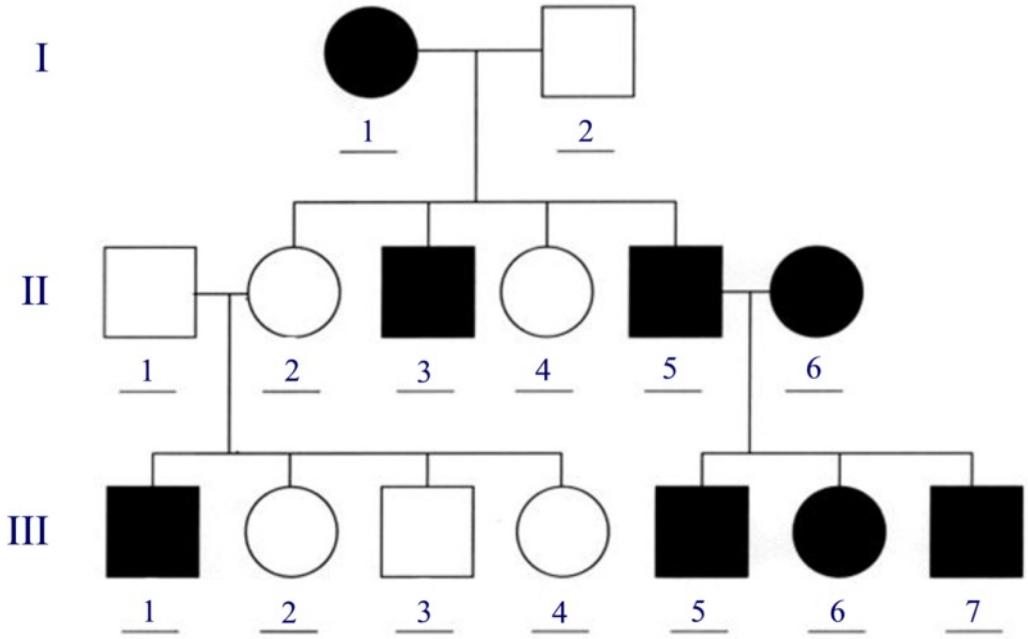
END OF SECTION A

END OF SECTION A

SECTION B – Short-answer questions

Question 1

The pedigree shows the inheritance of a condition in a family over several generations. Shading indicates expression of the condition.



a. What is the mode of inheritance?

1 mark

Solution

X-linked recessive

b. If III5 has a child with an individual who is heterozygous for the condition, what is the chance that they will have a child with the condition?

1 mark

Solution

50%

c. Using appropriate allele symbols, assign a genotype to Individual II2.

1 mark

Solution

X^AX^a where X^A – dominant allele for trait and X^a – recessive allele for trait

An ideogram of the chromosome that carries the gene for the inherited condition in the pedigree is shown below.

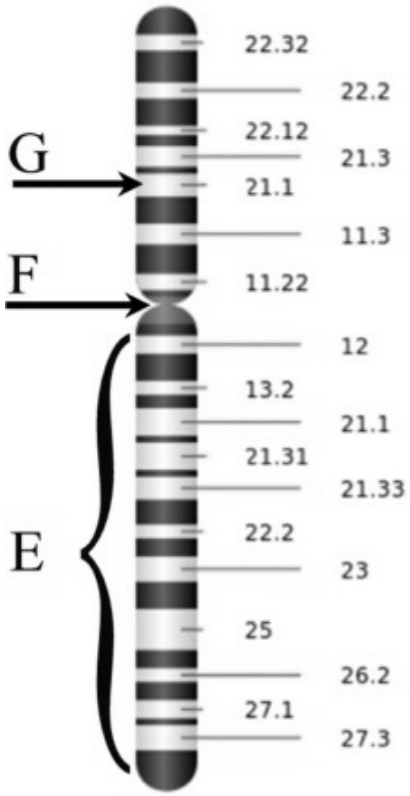


Image modified from http://en.wikipedia.org/wiki/File:Chromosome_X.svg

d. Complete the labelling of the diagram by filling in the table.

Structure	Name	Function
E	q – arm	
F		point of mitotic spindle attachment during cell division
G	locus	

2 marks

Solution

Structure	Name	Function
E	q – arm	**long arm of the chromosome**
F	**centromere**	point of mitotic spindle attachment during cell division
G	locus	**specific location of a gene or DNA sequence on a chromosome**

Mark allocation

- 2 marks if all 3 correct
- 1 mark if 2 or 1 correct
- 0 marks if 0 correct

e. How many copies of this chromosome would be present in a normal germline cell?

1 mark

Solution

If the germline cell is in a woman, there will be two of these chromosomes present OR if the germline cell is in a man, there will be one of these chromosomes present in a germline cell.

Note

Students do not need to provide both scenarios; only one is required and it must be correct to gain the mark.

Total 6 marks

Question 2

The following diagram outlines processes that occur within a living cell. The diagram is not drawn to scale.

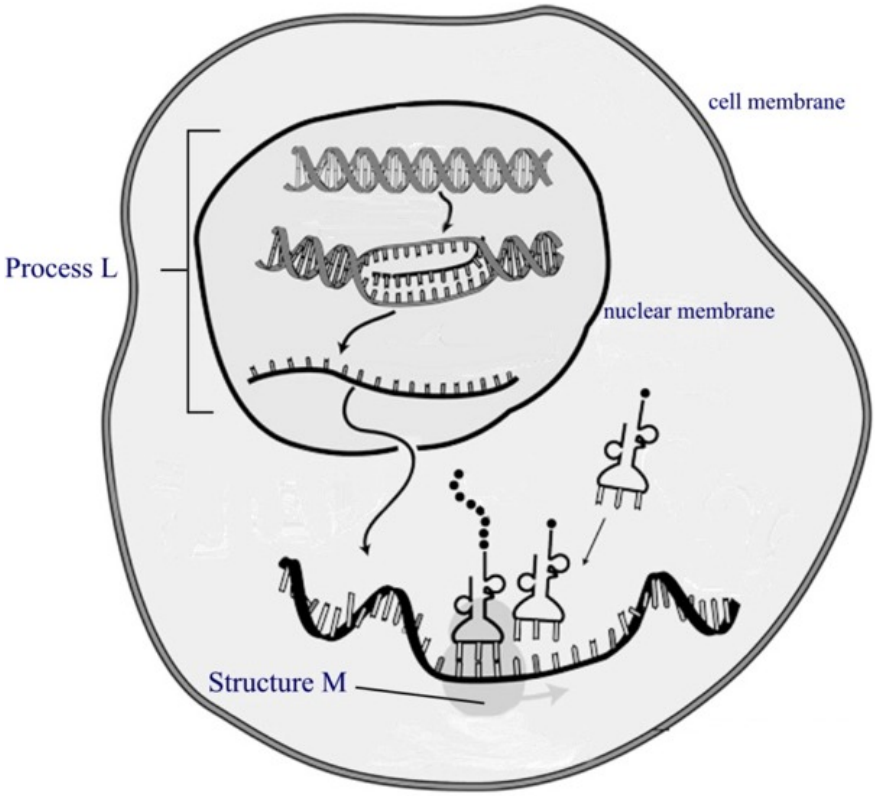


Image modified from http://en.wikipedia.org/wiki/File:Transkription_Translation_01.jpg

a. What is the name given to the structure in which helicase is found?

1 mark

Solution

The nucleus

b. Describe the function of helicase.

2 marks

Solution

Helicase is the enzyme that separates the strands of a DNA double helix by breaking the hydrogen bonds between nucleotide bases.

Mark allocation

- 1 mark – enzyme that separates the strands of a DNA double helix
- 1 mark – acts by breaking the hydrogen bonds between nucleotide bases

c. **i.** Identify the process that occurs at L.

1 mark

Solution

Transcription

ii. Describe the sequence of events that takes place during the process at L.

3 marks

Solution

The double-stranded DNA helix unzips, or opens, at the relevant position and the DNA template is used by RNA polymerase to synthesise a pre-messenger RNA (pre-mRNA) molecule. This undergoes post-transcriptional modification in which the introns are removed. Exons remain; mRNA is produced.

Mark allocation

- 1 mark – double stranded DNA unzips/opens, the DNA template is used by RNA polymerase to synthesise pre-mRNA
- 1 mark – this undergoes post-transcriptional modification in which the introns are removed
- 1 mark – exons remain, mRNA is produced

d. Name structure M. Explain how it contributes to protein production.

1 mark

Solution

Structure M: ribosome
The ribosome assembles amino acids to produce a specific protein molecule determined by the nucleotide sequence of an mRNA molecule.

Note

0 marks if student only identifies the structure or if contribution to protein synthesis is incorrect.

Total 8 marks

Question 3

Haemoglobin is the iron-containing protein in red blood cells that carries oxygen in the human body. The haemoglobin molecule (HbA) is made up of four polypeptide chains which consist of two alpha (α) globin chains and two beta (β) globin chains. Individuals with beta (β) thalassaemia do not produce β-globin and therefore cannot produce HbA. Mutations in the HBB gene cause β-thalassaemia.

Image from
http://en.wikipedia.org/wiki/File:1GZX_Haemoglobin.png

There are over 250 mutations in the HBB gene that are known to cause β-thalassaemia. One such mutation is directly associated with a condition known as sickle cell anaemia (SCA), in which an abnormal version of β-globin called haemoglobin S (HbS) replaces both β-globin chains in the haemoglobin molecule, causing the red blood cells to elongate, curve and take on a sickle shape. This change makes the sickled red blood cells less efficient at transporting oxygen through the bloodstream. The mutation that causes SCA is shown below.

HBB Sequence in Normal Adult Haemoglobin (HbA)

Nucleotide	CTG	ACT	CCT	GAG	GAG	AAG	TCT
Amino acid	Leu	Thr	Pro	Glu	Glu	Lys	Ser
	3			6			9

HBB Sequence in Mutant Adult Haemoglobin (HbS)

Nucleotide	CTG	ACT	CCT	GTG	GAG	AAG	TCT
Amino acid	Leu	Thr	Pro	Val	Glu	Lys	Ser
	3			6			9

a. Identify the mutation that causes SCA.

1 mark

Solution

Point mutation or substitution mutation or single base substitution

Malaria is a disease caused by a parasite of the genus *Plasmodium. Plasmodium* infects red blood cells and, whilst within, multiplies asexually. The parasite thrives in red blood cells that contain HbA, but is unable to survive in red blood cells containing HbS. However, individuals with red blood cells containing HbS have SCA. Consider the table below.

Genotype	Phenotype	Effect of genotypes	
		Malaria-affected environments	Malaria-free environments
HbA HbA	normal red blood cells haemoglobin A only	no sickling can die from malaria	no sickling
HbA HbS	red blood cells capable of sickling haemoglobin A and S	minor effects of sickling resistant to malaria	minor effects of sickling
HbS HbS	severe sickle cell anaemia haemoglobin S only	can die from sickle cell anaemia resistant to malaria	can die from sickle cell anaemia

b. Explain the observation that in areas with persistent malaria outbreaks, individuals with the HbA–HbS genotype occur in far greater numbers.

2 marks

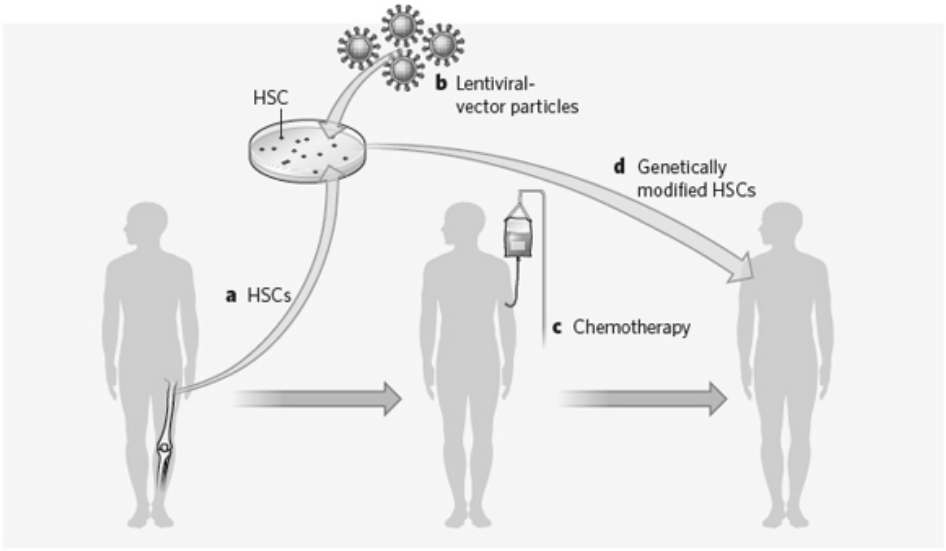
Solution

This is known as the heterozygote advantage. The HbA HbS genotype confers a higher relative fitness for individuals in malaria-affected environments. They do not die from SCA or malaria and so can survive to reproductive age.

Mark allocation

- 1 mark – heterozygote advantage – higher relative fitness in malaria-affected environments
- 1 mark – they do not die from SCA or malaria and so can survive to reproductive age

In 2007, an international team of scientists, led by biotherapist Marina Cavazzana-Calvo, took a sample of haematopoietic stem cells (HSCs) from the bone marrow of an 18-year-old man with β-thalassaemia who was dependent on blood transfusions for survival. The cells were cultured and mixed with lentiviral-vector particles into which a functional copy of the β-globin gene had been introduced. The genetically modified cells were transplanted. Levels of healthy red blood cells with normal β-globin chains gradually rose until, around a year after the treatment, he no longer required transfusions.



c. **i.** Why are HSCs so suitable for gene therapy?

1 mark

Solution

These cells can give rise to all blood cell types, including the haemoglobin-containing red blood cells, and can be used for a wide range of purposes.

ii. What would need to be guaranteed when using the viral vectors for the gene therapy?

1 mark

Solution

Ability to reproduce is removed OR only able to act as a vehicle for shuttling genes into cells OR that the virus is designed to inactivate itself after it transfers the modified genes OR virus does not make the patient sick.

Before receiving the genetically modified HSCs, the man was given chemotherapy.

d. Explain why chemotherapy was necessary at this stage of the procedure.

1 mark

Solution

To eliminate as many of the faulty HSCs as possible so that dilution of the genetically corrected cells would not occur.

The treatment has been viewed as successful; however the scientific community remains cautious. Concerns have been raised about overexpression of a protein called HMGA2 which resulted in a high proportion of the genetically modified cells. HMGA2 has been linked to cancers.

e. **i.** What event might have triggered overexpression of HMGA2?

1 mark

Solution

The lentivirus vector could have randomly integrated into the chromosome which contained the HMGA2 gene leading to the rapid proliferation of the cells.

ii. If future treatments are to proceed, how might overexpression of HMGA2 be avoided?

1 mark

Solution

By extracting a larger initial population of modified HSCs, researchers could potentially prevent the problem from occurring.

Total 8 marks

Question 4

The Komodo dragon (*Varanus komodoensis)* has the ZW chromosomal sex-determination system. Female dragons carry one Z and one W chromosome and males carry two Z chromosomes. In 2005 at London Zoo, after being separated from a male Komodo dragon for more than two years, a female dragon (Sungai) laid a clutch of eggs. Scientists erroneously believed that she had been able to store sperm from an earlier encounter. In 2006, it was reported that another Komodo dragon (Flora) at a different zoo, laid a clutch of unfertilised eggs. Of the 11 eggs, seven hatched, all of them male.

Genetic tests were carried out on three eggs that collapsed after being moved to an incubator. The results reported that the offspring were not identical clones.

a. What is the term used to describe the female Komodo dragon genotype?

1 mark

Solution

heterogametic

b. Explain whether it is possible for all offspring to be homozygous at all loci.

2 marks

Solution

No it is not, the statement implies that there was no crossing over at all in meiosis I. There should be crossing over which produces *reduced* heterozygosity but not complete homozygosity.

Mark allocation

- 1 mark – no, the statement ignores the possibility of crossing over in meiosis I
- 1 mark – crossing over will occur, resulting in *reduced* heterozygosity

c. Draw a diagram to clearly demonstrate the mechanism that produces unfertilised eggs that develop into male Komodo dragons. Include written explanations for clarity.

2 marks

Solution

Diagram should show that in the process of meiosis one cleavage (most likely the second meiotic cleavage) was suppressed, so both homologous chromosomes in the resultant ovum were derived from the same replicated DNA strand.

Mark allocation

- 1 mark – correct process of meiosis to the first cleavage, showing double stranded homologous chromosomes pairing up
- 1 mark – suppression of second cleavage, showing no further cleavage and retention of double-stranded chromosomes

One advantage of this reproductive strategy is that Komodo females can reproduce without male mates. In the wild, a female Komodo dragon could swim to a new island, lay a clutch of unfertilised eggs and potentially found an entirely new colony on its own.

d. What is the disadvantage in parthenogenetic reproduction in Komodo dragons?

2 marks

Solution

All hatchlings will be males; they would interbreed with their own mother and within one generation, there would potentially be a population able to reproduce normally on the new island. The entire colony would have very low genetic diversity leading to an inbreeding depression and low genetic fitness.

Mark allocation

- 1 mark – they would interbreed with their own mother; within one generation, there would potentially be a population able to sexually reproduce on the new island
- 1 mark – the entire colony would have very low genetic diversity leading to an inbreeding depression and low genetic fitness

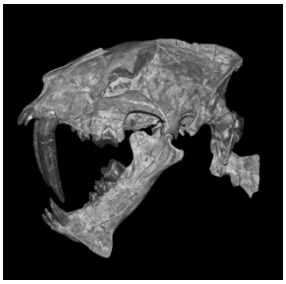
Total 7 marks

Question 5

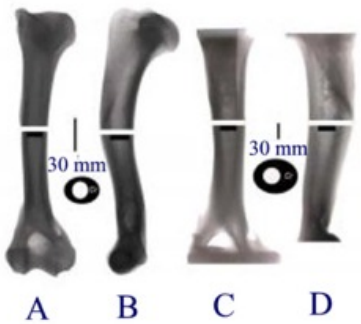
The sabre-toothed cat *Smilodon* (2 mya–1 mya) has been described as a 'hyper-ambush' hunter. Instead of using the ambush–chase strategy seen in modern cats, it is believed *Smilodon* would hide and lie in wait for its prey. When prey did pass by, *Smilodon* would leap out at it, probably knocking it off balance and attacking.

Unlike the modern cat which imparts death by using its jaws to close off the throat or nose of its prey, the sabre-toothed cat used its teeth to inflict quick lethal stabs. The teeth of the modern cat are conical and able to withstand forces in all directions. Sabre teeth were long and flattened and if they struck the bones of prey during a bite would have shattered very easily.

Studies of fossils suggest that at least three different groups of ancient animals hunted in the same way. The following table presents information about four groups of 'hyper-ambush' hunters.

Smilodon fatalis (sabre-tooth cat)	***Hoplophoneus primaevus*** Nimravidae	***Barbourofelis loveorum*** Barbourofelidae	***Panthera atrox*** (extinct American lion)
North America South America	North America Eurasia	North America Eurasia Africa	North America
2 mya–1 mya	42 mya–7 mya	16 mya–9 mya	0.34 mya–11,000 ya
 *Image: Stickpen*	 *Image: H Zell*	 *Image: Dallas Krentzel*	 *Image: Claire H*

X-rays comparing the cross-sectional dimensions of the upper arm bone of a jaguar (A and B) with *Smilodon fatalis* (C and D) showed that the upper arms had extra-thick, reinforced bone with significantly large attachment points.



a. Explain the significance of the upper arm bones and the implications for the teeth of the sabre-toothed cat.

2 marks

Solution

Reinforced bone was to provide extra bulk and a foundation for the large attachment points to which muscles would have been attached, giving the sabre-toothed cat strong and powerful arms for holding onto prey during the kill. This enabled preservation of the sabre teeth which could have been damaged during the tussle associated with the killing.

Mark allocation

- 1 mark – to provide extra bulk and a foundation for the large attachment points to which muscles would have been attached, giving the sabre-toothed cat strong and powerful arms for holding onto prey whilst it was killed
- 1 mark – this enabled preservation of the sabre teeth which would have been used for the killing

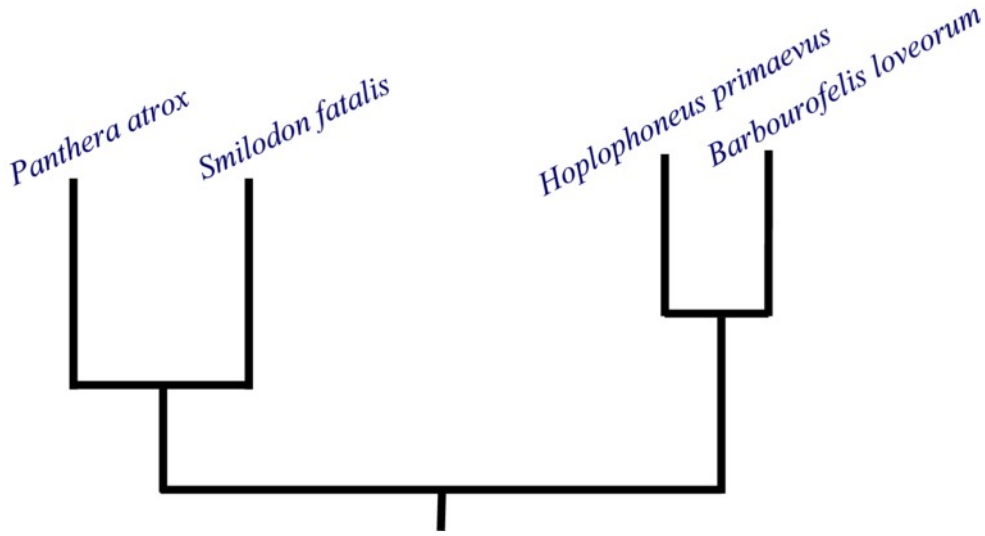
b. What prediction could be made about the relationship between tooth length and the size and shape of forelimb bones?

1 mark

Solution

The longer the teeth, the sturdier the forelimb bones and, probably, the wider the paws.

The cladogram shows the evolutionary relationship that exists between the four groups of 'hyper-ambush' hunters. They are separated by considerable distance in geological time.



c. Given the strong similarities shown by each of these groups, identify and describe what kind of evolution has taken place.

2 marks

Solution

Convergent evolution - the species are not related by a recent common ancestor. Similar characteristics (size and shape of forelimbs, teeth, skulls and general physique) have developed in unrelated species owing to the similar ecological niches occupied by the animals.

Mark allocation

- 1 mark – convergent evolution - species are not related by a recent common ancestor
- 1 mark – similar characteristics have developed in unrelated species due to the similar ecological niches occupied by the animals

In many regions around the globe, 'hyper-ambush' hunters are known to have hunted the same prey as their shorter-toothed relatives.

d. Suggest two strategies the 'hyper-ambush' hunters might have used in order to reduce competition and live side by side with their shorter-toothed relatives.

2 marks

Solution

They use a different hunting technique and live in slightly different habitats.

Mark allocation

- 1 mark – use of different hunting technique
- 1 mark – live in slightly different habitats

Total 8 marks

Question 6

The common monkey-flower *Mimulus guttatus* is an annual (lives for one year) or perennial (lives for two years) herbaceous wildflower that has a very broad geographical and ecological range in western North America.

	Ecotype	Habitat	Flowering time
Mimulus guttatus (AN)	annual	dry inland	early in the season
Mimulus guttatus (PE)	perennial	moist and cool coastal	late in the season

a. Explain the likely effect of the different flowering times on the two ecotypes of *Mimulus guttatus*.

1 mark

Solution

Pre-zygotic isolation is the likely effect due to the fact that the two ecotypes do not flower at the same time and cross pollination cannot occur.

The diagram shows chromosome AN and chromosome PE. The chromosomes carry the same genes.

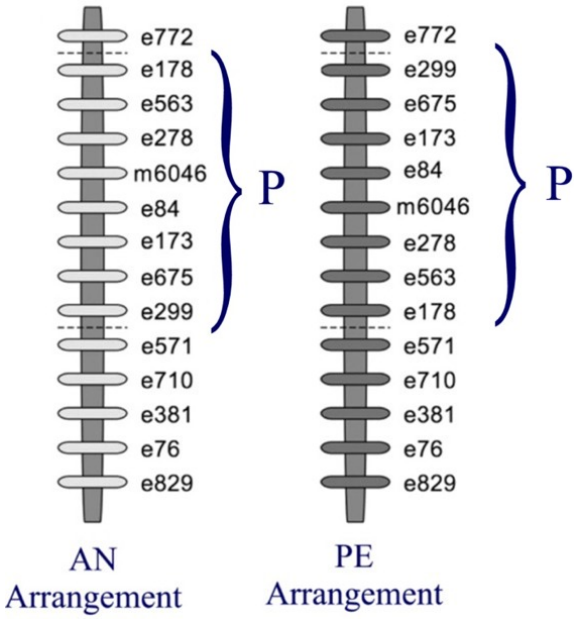


Image: Lowry DB, Willis JH (2010) A Widespread Chromosomal Inversion Polymorphism Contributes to a Major Life-History Transition, Local Adaptation, and Reproductive Isolation.

b. Suggest a likely explanation for the appearance of these two chromosomes and state what event could have caused this to occur.

2 marks

Solution

A chromosomal inversion has occurred. A segment of DNA has been cut out of the chromosome and then reinserted upside down.

Mark allocation

- 1 mark – a chromosomal inversion
- 1 mark – a segment of DNA has been cut out of the chromosome and reinserted upside down

David Lowry, the researcher investigating the monkey-flower, found a large suite of 350 adaptive genes within the section shown at P. These genes determine whether the plant will survive in an inland or a coastal habitat. The adaptive genes of the inland and the coastal monkey-flower appear to be 'locked-in' position. If transplanted to the other environment, neither variety is able to reproduce successfully. The consequence of its spread is reproductive isolation and potentially a new species.

c. i. Explain the mechanism resulting in the reproductive isolation of *M. guttatus* (AN) and *M. guttatus* (PE).

2 marks

Solution

The inversion of 350 genes will render the chromosomes effectively non-homologous and gene expression will no longer be normal.

Mark allocation

- 1 mark – chromosomes become effectively non-homologous
- 1 mark – gene expression is no longer normal

ii. What form of evolution is taking place within the *M. guttatus* population?

1 mark

Solution

Divergent evolution

Total 6 marks

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Question 7

The genus of plants *Penstemon* is endemic to the Great Basin region of the western United States.

Populations of *Penstemon* are found living at high elevation on separate mountain tops. The seeds are all dispersed by gravity and do not move very far from the parent plant.

a. Suggest how gene flow is most likely to occur between these populations.

1 mark

Solution

Gene flow will occur through the movement of pollen between populations.

Penstemons from 6 to 8 populations on 4 to 6 mountain ranges were sampled. By extracting DNA from leaf tissues up to 8 polymorphic microsatellite loci were identified.

b. What is a polymorphic microsatellite?

2 marks

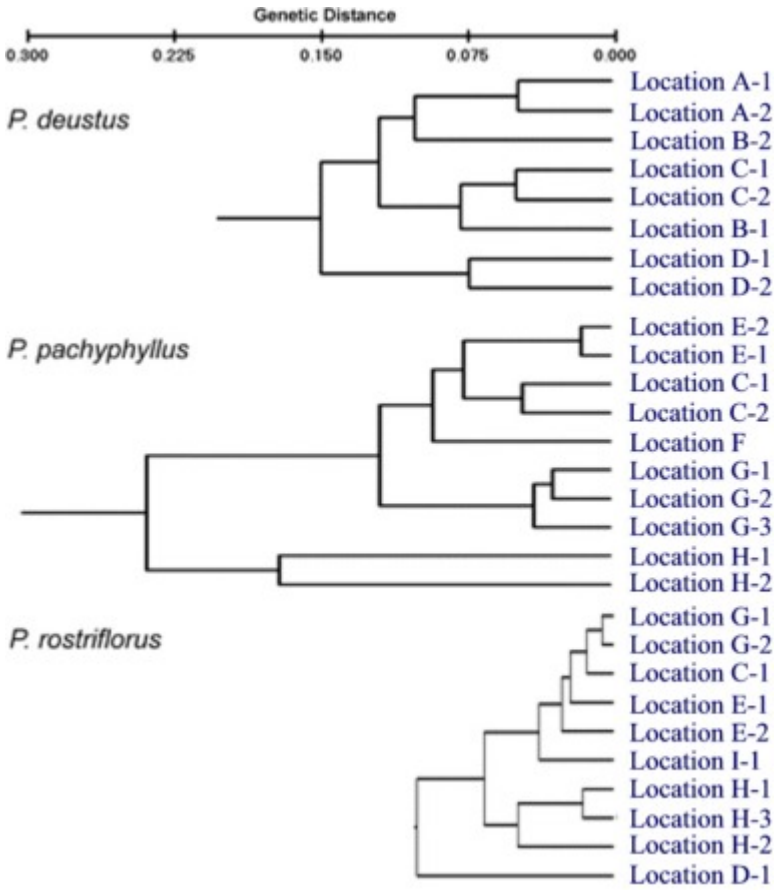
Solution

A microsatellite is a small sequence of DNA that is repeated many times. When the number of repeats for the particular sequence varies between organisms, a microsatellite is called polymorphic.

Mark allocation

- 1 mark – microsatellite is a small sequence of DNA that is repeated many times
- 1 mark – polymorphic – when the number of repeats varies between organisms

This data was then used to determine patterns of gene diversity both within each population on a mountain top and between more distant populations found on other mountain tops. The degree of genetic distance between the different populations is shown below.



c. In relation to the cladogram, what can be observed about the genetic distance that exists in the populations of *P. pachyphyllus* and *P. rostriflorus*?

1 mark

Solution

The genetic distance that exists within the ten populations of *P. pachyphyllus* is double the genetic distance that exists amongst the ten populations of *P. rostriflorus*.

The table shows three species of *Penstemon* and their mode of pollination.

Species of *Penstemon*	Mode of pollination
P. pachyphyllus, P. deustus	bees
P. rostriflorus	hummingbirds

d. Suggest an explanation for the genetic distance observed in *P. rostriflorus*.

1 mark

Solution

P. rostriflorus is pollinated by hummingbirds which may more easily be able to travel the distance between the different mountain top populations, particularly in comparison with bees.

The study showed that bee-pollinated species of *Penstemon* were genetically clustered and distinct for each mountain range. There was little or no mixing of genetic material between mountain ranges.

e. Clearly outline the stages of the process that would have resulted in the genetic isolation of *P. pachyphyllus* and *P. deustus*.

3 marks

Solution

Originally one population became separated thus halting gene flow. Natural selection occurred and when the populations were brought back together they were unable to produce viable/fertile offspring.

Mark allocation

- 1 mark – one population became separated thus halting gene flow
- 1 mark – natural selection occur
- 1 mark – when brought back together they were unable to produce viable/fertile offspring

Total 8 marks

END OF SOLUTIONS BOOK