

OCR A-LEVEL BIOLOGY B • H422

BIOLOGY



EXAM QUESTION PACK

5.1.2 POPULATION GENETICS AND EPIGENETICS

SCORE

/98

PERCENTAGE

%

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- 1 Cambodia is thought to have the largest wild population of the Siamese crocodile, *Crocodylus siamensis*.

C.siamensis is a critically endangered species that is now virtually extinct in the wild in most other countries of Southeast Asia.

Studies of genetic diversity amongst the Cambodian population of *C.siamensis* in protected areas have provided some evidence that there was a recent genetic bottleneck event in the Tonle Sap Lake area.

- (i) Using this example, explain what is meant by the term, genetic bottleneck.

[3]

- (ii) Give **one** possible cause for the genetic bottleneck event that affected *C.siamensis*.

[1]

2(a)

The following passage describes aspects of epigenetics.

Complete the passage using the most appropriate terms.

A mutation causes a change in the nucleotide sequence of DNA. Epigenetic changes do not change the nucleotide sequence. Instead, they change the extent to which genes are

One epigenetic mechanism involves acetylation of, which makes the genes more accessible. Another mechanism involves of the DNA. Some epigenetic changes can be inherited if they are present in [4]

(b) Twenty zebrafish were exposed to hypoxia (oxygen deprivation) by keeping them in water with a low oxygen concentration for 4 weeks.

All the offspring of these fish had greater resistance to hypoxia than offspring whose parents were kept in well-oxygenated water.

The researchers concluded that this showed evidence for inheritance of epigenetic changes rather than being the result of a mutation.

Evaluate this conclusion.

----- [3]

(c) *Drosophila simulans* and *Drosophila mauritiana* are two closely related species of fruit fly.

The *OdsH* gene in *Drosophila* is described as a speciation gene. It causes epigenetic changes to the DNA that regulates the condensation of chromosomes.

(i) Name the stage of the cell cycle in which condensation of chromosomes occurs.

-----[1]

(ii) The *OdsH* gene causes male sterility in *D. simulans* – *D. mauritiana* hybrids.

Based on the information provided, suggest how the *OdsH* gene has caused *D. simulans* and *D. mauritiana* to become and remain separate species.

-----[3]

- 3 Please refer to the Advance Notice article 'Do Pathogens Gain Virulence as Hosts Become More Resistant?' in Insert H422-02 October 2021.

The article describes the interactions between pathogen virulence and host resistance to infection.

- (i) Explain how immunological and genetic factors would allow a population of wild rabbits to become more resistant to infection by MYXV.

[2]

- (ii) The article discusses how resistance to infection can be affected by two common agricultural practices: vaccination and selective breeding.

Explain how vaccination is used to protect poultry against pathogens such as Marek's disease virus (MDV).

[3]

- (iii) Explain why vaccination might **not** prevent epidemics of Marek's disease in poultry.

[2]

(iv) Evaluate the suggestion that birds should be bred for susceptibility to MDV rather than resistance.

[4]

- 4 Please refer to the Advance Notice article 'Learning from Iceland's model for genetic research' in the Insert for QP H422/02, June 19.

The article explains how Iceland has one of the most genetically homogeneous populations in Europe.

(i) State what is meant by the following genetic terms.

1. Gene -----

2. Allele -----

[2]

(ii) Allele frequencies will remain constant in a stable population. However, population changes can alter allele frequencies.

For each of the following events, give the term used to describe the effect on allele frequencies.

1. Iceland was settled by a relatively small number of explorers in the 9th century.

2. The Icelandic population decreased due to the two waves of plagues during the 15th century, smallpox in the early 1700s and a volcanic eruption in 1783.

[2]

(iii) Explain how the events described in (ii) resulted in the low genetic diversity of the Icelandic population.

[2]

5 Which of the statements, **A** to **D**, is true of epigenetics?

- A** Guanine is the most commonly methylated DNA base.
- B** Identical twins show identical epigenetics.
- C** Proteins cannot undergo epigenetic modification.
- D** Some epigenetic changes can be reversed.

Your answer

[1]

6 Freeman was working on developing drought-resistant varieties of alfalfa using selective breeding, but this has proved difficult.

Drought resistance depends on the ability to withstand several abiotic factors, such as high temperatures and high light intensity.

- (i) Use your knowledge of inheritance to suggest why it is difficult to study the genetic basis of drought resistance.

----- [2]

- (ii) Alleles of the *miRNA* 156 gene regulate a group of transcription factors in alfalfa. These transcription factors activate or inhibit promoters that control genes related to drought resistance.

Explain how the *miRNA* 156 gene could be used to investigate the genetic basis of drought resistance.

----- [2]

- (iii) Scientists have made a plasmid that produces more of the *miRNA* 156 gene product than normal and want to use this to develop a drought-resistant alfalfa plant.

Explain how they could incorporate the plasmid into alfalfa cells.

----- [2]

7 The gene, *HBB*, codes for the beta polypeptide in haemoglobin.

A person with sickle cell anaemia has a mutation in *HBB*. This causes a change to the sixth amino acid in the beta polypeptide.



Scientists carried out a study to investigate the effect of the sickle cell allele on the survival rate of children in Kenya.

Data were collected using questionnaires completed by health workers visiting families with young children. The questionnaires were collected from health centres in both urban and rural parts of Kenya.

The data were analysed and 867 children were grouped based on their *HBB* genotypes, which the health centres supplied, as shown in Table 31.

<i>HBB</i> genotype	Number of children
H ^A H ^A	392
H ^A H ^S	374
H ^S H ^S	101

Table 31

H^A is the healthy allele and H^S is the sickle cell allele.

The percentage of children surviving with each *HBB* genotype was recorded from birth at 60-day intervals until day 600.

Fig. 31 shows the data from this study.

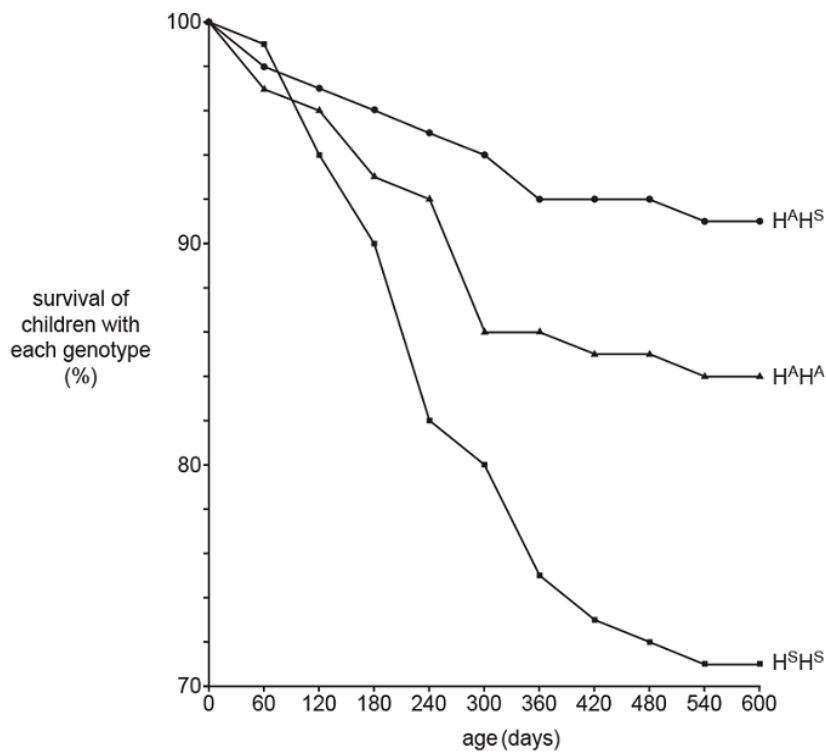


Fig. 31

Malaria is a leading cause of mortality in Kenya.

Using this information and your own knowledge, describe and explain the data in Fig. 31 and comment on its quality.

[6]

8 The statements below relate to the epigenetic regulation of gene expression.

Which of the following statements is/are correct?

- 1 Methylation of DNA prevents gene transcription.
 - 2 The most common base to undergo methylation is guanine.
 - 3 Acetylation of histone proteins causes DNA to become less accessible to transcription factors.
- A 1, 2 and 3 are correct
- B Only 1 and 2 are correct
- C Only 2 and 3 are correct
- D Only 1 is correct

Your answer

[1]

- 9 Diseases that involve chronic inflammation, such as asthma, are treated with steroids. Steroids reduce the transcription of the genes coding for molecules that produce an inflammatory response.

Steroids affect structural cells and cells involved in the inflammatory response. Structural cells form the tissues of the respiratory system.

- (i) Suggest what effect steroids have on histones such that gene transcription is reduced.

----- [1]

- (ii) Steroids can suppress the production of certain molecules by cells in the respiratory system.

Suggest one molecule released by a cell involved in an inflammatory response **and** one released by a structural cell for which production is suppressed by steroids.

molecule in inflammatory response -----

molecule released by structural cell -----

[2]

- (iii) Acute asthma attacks are treated using beta-agonists.

Name the tissue targeted by beta-agonists **and** state the location of this tissue in the airways.

tissue -----

location of tissue -----

[2]

In an island population, 1 in 12 500 individuals were found to have low levels of catalase activity in erythrocytes. Individuals with low levels of erythrocyte catalase activity have no related health problems.

- (i) Calculate the frequency of the normal allele for the catalase gene within this population using the equation below. You should assume the gene has only two alleles.

$$p + q = 1$$

Show your working. Give your answer to **three** significant figures.

Answer [2]

- (ii) In other parts of the world, the frequency of individuals with low erythrocyte catalase activity is between 1 in 20 000 and 1 in 25 000.

What is the most likely explanation for the difference in frequency in the island population given in (i)?

.....
.....
..... [1]

11 The frequency of the sickle cell allele in human populations has been found to be higher in areas of the world such as India and the west coast of Africa. In these regions, malaria is said to be endemic.

(i) State what is meant by the term *endemic*.

----- [1]

(ii) Explain why the frequency of the sickle cell allele is higher in areas such as India and the west coast of Africa.

----- [4]

- 13 An individual who experiences poor nutrition as a fetus has a higher probability of developing diabetes in adulthood.

Suggest the type of diabetes that is likely to develop in adults as a result of fetal under-nutrition.

Explain your choice.

[3]

14(a) In human DNA, the frequency of CpG sites across the genome varies.

In the human genome:

- analysis of the base content of human DNA shows that 21% of the bases present are cytosine.
- the **overall** frequency of CpG sites in the human genome is 0.01.
- in regions of the genome known as **promoters**, the frequency of CpGs is approximately 0.6.
- deamination of cytosine at CpG sites converts the cytosine to **thymine**.

(i) Using the information about the human genome, calculate the **expected** frequency of a CpG site in human DNA.

Show your working. Give your answer to **three** significant figures.

Answer = _____ [2]

(ii) By analysing the information about the human genome and your answer to (i), explain why the frequency of CpG sites is low in some regions but high in others.

[4]

- (b) In the Advanced Notice article you were told that HDAC and DNMT inhibitors are now available and used as anticancer drugs.

Fig.1.4 shows a section of DNA and its associated histone proteins.

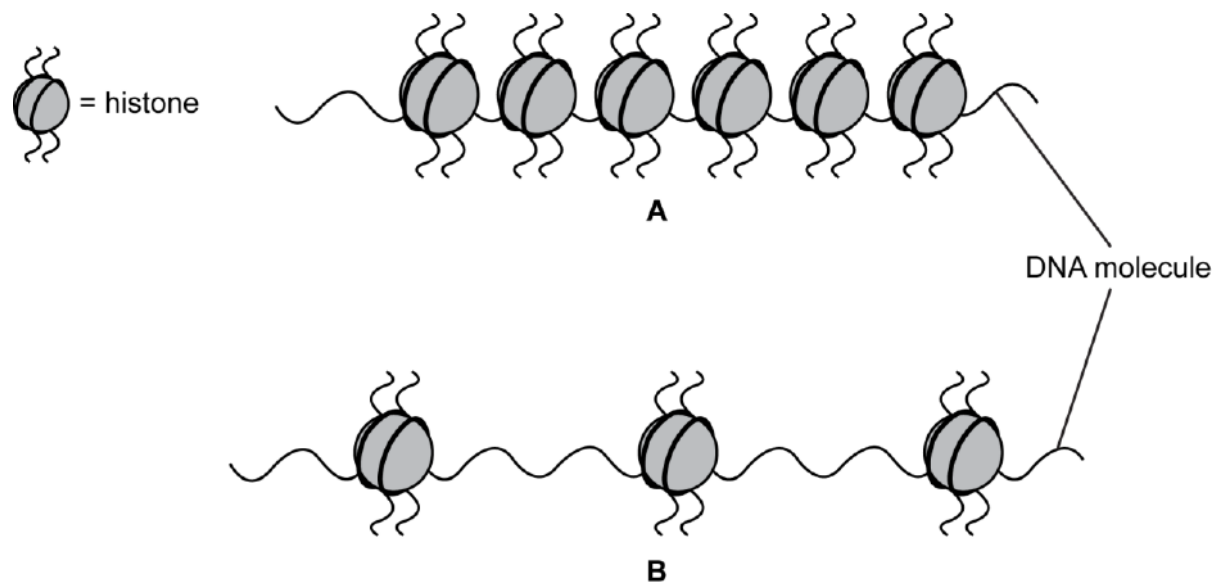


Fig. 1.4

- (i) Which of the two diagrams in Fig.1.4, **A** or **B**, shows the DNA and histones from a cancer cell that has been successfully treated with an HDAC inhibitor? Explain your answer.

[2]

- (ii) Inactivation of DNMT during DNA replication leads to the **loss** of methyl groups from DNA in dividing cancer cells.

Suggest why.

[1]

15 The Hh blood group system is controlled by one gene locus with two alleles.

The homozygous recessive genotype produces the Bombay phenotype, resulting in a very rare blood group, in which no antigen is expressed.

The Bombay phenotype is very rare. One person in 250 000 of the world's population is estimated to have the Bombay phenotype.

- (i) Using the Hardy-Weinberg equations, calculate the **percentage** of the world's population who carry one copy of the recessive allele.

$$p + q = 1 \quad p^2 + 2pq + q^2 = 1$$

Show each step in your working. Give your answer to **one significant figure**.

percentage [4]

- (ii) The Bombay phenotype is more common in some regions of India, where it can occur in one in 10000 people.

Researchers have suggested that the Bombay phenotype is more common in these regions because of the practice of endogamy, in which marriage occurs only between people within the same tribe or small social group.

Suggest why endogamy has increased the frequency of the Bombay phenotype.

.....
.....
.....
.....
..... [2]

- 16 People travelling to high altitudes can develop altitude sickness because they produce more haemoglobin than normal, which results in thick, viscous blood.

Many people in Tibet live more than 4 000 m above sea level, but they do not develop altitude sickness.

Tibetan people have a variant of the EPAS1 gene that causes them to maintain relatively low haemoglobin levels in their blood.

- (i) Describe how the Tibetan variant of the EPAS1 gene has become common in Tibetan populations.

[4]

- (ii) What type of adaptation is represented by the maintenance of low haemoglobin levels in Tibetan people?

[1]

- (iii) State and explain **one** problem that a Tibetan person with the EPAS1 gene variant might experience.

[2]

- (iv) Suggest a practical technique that could be used to compare the relative number of erythrocytes in a Tibetan population with that of another population **and** state what this technique would show.

[2]

END OF QUESTION PAPER

Mark Scheme

Question			Answer/Indicative content	Marks	Guidance
1		i	chance event occurs ✓ (causing) <u>significant</u> reduction in , population / number of individuals , in Tonle Sap Lake area ✓ surviving crocodile population have fewer alleles than original population ✓ decrease in genetic diversity ✓	3 max(A O2.1)	
		ii	habitat destruction ✓ over-hunting / over-poaching ✓ disease ✓ toxic chemicals in the lake ✓ loss of prey species ✓ named natural disaster ✓	1 max	ALLOW description Must be relevant to the lake e.g. drought IGNORE flooding
			Total	4	
2	a		transcribed / expressed ✓ histones ✓ methylation ✓ gametes ✓	4(AO1.1) (AO1.1)(AO1.1)(A O1.1)	ALLOW translated ALLOW (named) sex cells
	b		mutation is unlikely to , have occurred in many parents / affect all offspring ✓ (whereas) DNA methylation / acetylation of histones , could have occurred in, many / all , parents (exposed to hypoxia) ✓ (which) activates / inhibits , expression of genes related to resistance to hypoxia ✓ lack of oxygen is a selective pressure ✓ (but biochemical) evidence of epigenetics in offspring is needed ✓	max 3(A O3.2)(AO 3.2)(AO3. 2)(AO3.2) (AO3.2)	
	c	i	prophase ✓	1	ALLOW mitosis

Mark Scheme

Question			Answer/Indicative content	Marks	Guidance
		ii	(reference to) reproductive isolation ✓ epigenetic changes alter gene expression ✓ stops , meiosis / gamete formation ✓ (because) hybrids are <u>infertile</u> ✓ (so) species would diverge / remain separate ✓	max 3(AO3.2)(AO3.2)(AO3.2)(AO3.2)(AO3.2)	
			Total	11	

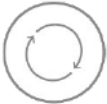
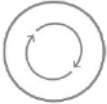
Mark Scheme

Question			Answer/Indicative content	Marks	Guidance
3		i	(surviving rabbits produce) memory (T / B) cells ✓ <u>alleles</u> for resistance (to MYXV) would be selected for ✓ (so the) <u>allele frequency</u> increases (in the population) ✓	max 2(AO2.2)(AO2.2)(AO2.2)	ALLOW advantageous allele(s) passed on
		ii	(vaccination involves) giving pathogen antigens ✓ (formation of) memory (T / B) cells ✓ exposure to pathogen leads to secondary immune response ✓ (which) destroys / kills, pathogen before it can cause, disease / death ✓	max 3(AO1.2)(AO1.2)(AO1.2)	ALLOW any correct example of vaccine, e.g. live-attenuated, subunit vaccines, pathogen fragments ALLOW rapid production of antibodies
		iii	vaccinated hosts can still transmit virus ✓ (so) unvaccinated, birds / flocks, could become infected ✓ OR (high) mutation rate leads to antigen variability ✓ so antibodies produced by vaccination no longer specific ✓	2(AO2.5)(AO2.5)(AO2.5)(AO2.5)	

Mark Scheme

Question			Answer/Indicative content	Marks	Guidance
		iv	<i>for</i> susceptible birds would die before they could infect other birds ✓ resistant birds (are more likely to) encourage evolution of more virulent pathogens ✓ <i>against</i> (but) evolution of virulence may not matter for resistant birds ✓ hypervirulent pathogens could threaten, non-resistant / non-GMO, flocks ✓ ethical issues if industrial practices threaten organic producers ✓ susceptible birds may also be susceptible to, related / similar, pathogens	max 4(AO3.2)(AO3.2)(AO3.2)(AO3.2)(AO3.2)	ALLOW other unambiguous term for birds throughout
			Total	11	

Mark Scheme

Question			Answer/Indicative content	Marks	Guidance
4		i	1 sequence of, DNA / nucleotides / bases, that code for a polypeptide ✓ 2 a gene variant ✓	2 AO1.1	Mark the first answer. If an additional answer is given that is incorrect, then = 0 marks ALLOW protein / amino acid sequence for 'polypeptide' ALLOW different versions of the same gene <u>Examiner's Comments</u> Most candidates could define allele and all candidates attempted the definitions. Gene descriptions were often given to GCSE standard with the use of 'coding for a characteristic' rather than protein.  AFL It is useful to tabulate or highlight key points that are extended at A Level compared to the candidate's GCSE knowledge/ specification.
		ii	1 <u>founder(s)</u> effect ✓ 2 <u>genetic bottleneck</u> ✓	2 AO2.1	Mark the first answer. If an additional answer is given that is incorrect, then = 0 marks <u>Examiner's Comments</u> This was a well answered question by candidates who could clearly identify the relevant terms in relation to the example given. A few candidates simply stated bottleneck for example 2.  AFL Splitting key terms to see their origins often helps to consolidate knowledge and avoid candidates attempting to summarise terms that then give a different meaning. Placing words separately into a search engine is a quick way of allowing students to suggest definitions of key terms they have not previously encountered.

Mark Scheme

Question			Answer/Indicative content	Marks	Guidance
		iii	1 small / decreasing , gene pool ✓ 2 limited number / loss, of <u>alleles</u> ✓	2 AO2.1	<u>Examiner's Comments</u> Most candidates recognised that alleles would be lost and limited in these circumstances and some candidates related that to the size of the gene pool in the population. Answers often referred to inbreeding but did not connect this to limited allele number.
			Total	0	
5			D	1	<u>Examiner's Comments</u> Only the more able candidates were able to select D as the correct response.
			Total	1	

Mark Scheme

Question			Answer/Indicative content	Marks	Guidance
6		i	(drought resistance) involves many, genes / loci ✓ Involves (possible), multiple / variety, alleles (at each locus) ✓	2	ALLOW (drought resistance) is polygenic IGNORE different alleles <u>Examiner's Comments</u> Some candidates were able to appreciate the polygenic aspect of drought resistance and successfully linked the information in the stem of the question with genes and inheritance. Many candidates did not recognise the relevance of 'inheritance' and 'genetic basis' in the stem of the question and discussed their answers in terms of phenotype only e.g. not many plants survive, so there are very few that can be studied.
		ii	(gene/ transcription factor) can be used to identify, genes / alleles, involved / activated, in drought resistance ✓ promoters allow, gene expression / transcription ✓ remove / knockout, (miRNA 156) gene and observe, phenotype / drought resistance ✓	2	ALLOW RNA Polymerase binds to promoters <u>Examiner's Comments</u> Candidates struggled to understand the relevance of the miRNA gene and confused this with RNA. However, many candidates did discuss knocking out miRNA to observe the phenotype and so exhibited a good understanding of knockout technology.

Mark Scheme

Question			Answer/Indicative content	Marks	Guidance
		iii	place plasmid in <i>Agrobacterium</i> / use gene gun ✓ screen for, transgenic/ recombinant, cells ✓ details of selectable marker ✓	2 max	ALLOW use <i>Agrobacterium</i> as vector for 'place plasmid in <i>Agrobacterium</i>'. ALLOW a description of a gene gun IGNORE ref to viruses / liposomes IGNORE ref to antibiotics <u>Examiner's Comments</u> Few candidates scored full marks. Those that scored one mark usually referred to a gene gun for insertion of the plasmid. Most candidates did not appreciate that the alfalfa would be insertion into plant cells and referred to vectors relevant to animal cells e.g. viruses. There were a lot of descriptions using restriction enzymes and ligases to obtain the recombinant plasmid, not realising the focus of the question was on the incorporation of the plasmid.
			Total	6	

Mark Scheme

Question	Answer/Indicative content	Marks	Guidance
7	Summary of instructions to markers: <i>Read through the whole answer. (Be prepared to recognise and credit unexpected approaches where they show relevance.) Using a 'best-fit' approach based on the science content of the answer, first decide which of the level descriptors, Level 1, Level 2 or Level 3, best describes the overall quality of the answer.</i> <i>Then, award the higher or lower mark within the level, according to the Communication Statement (shown in italics):</i> <i>o award the higher mark where the Communication Statement has been met.</i> <i>o award the lower mark where aspects of the Communication Statement have been missed.</i> <i>The science content determines the level.</i> <i>The Communication Statement determines the mark within a level.</i> Level 3 (5 – 6 marks) A detailed description and explanation of the data, comparing the survival of the <i>HBB</i> genotypes. There are some valid comments relating to data quality. <i>There is a well-developed line of reasoning which is clear and logically structured. The information presented is relevant and substantiated.</i> Level 2 (3 – 4 marks) Good description of the data, comparing the survival of the <i>HBB</i> genotypes. There is some explanation of survival of at least one genotype OR there is some valid comment on data quality. <i>There is a line of reasoning presented with some structure. The information presented is in the most-part relevant and supported by some evidence.</i>	6	Indicative scientific points may include: H^SH^S genotype - fastest death rate / most deadly genotype - ref to haemoglobin / erythrocyte clumping - ref to consequent capillary-blocking & organ damage H^AH^S genotype - lowest death rate / least deadly genotype - due to lower risk of malaria - least infectivity of / best resistance to malaria parasite (<i>Plasmodium</i>) - ref to sickling of erythrocytes in low oxygen tension and destruction of parasite within by phagocytosis

Mark Scheme

Question	Answer/Indicative content	Marks	Guidance
	Level 1 (1 – 2 marks) Good description of the data, comparing the survival of the <i>HBB</i> genotypes OR limited explanation of the data. There is no comment on data quality. <i>There is an attempt at a logical structure with a line of reasoning. The information is in the most part relevant.</i> 0 marks No response or no response worthy of credit.		- other detail, e.g. ref to peroxide in sickled cells killing parasite and ref to increase in carbon monoxide production as possible reason for lower risk of malaria H^AH^A genotype - death rate between H^SH^S and H^AH^S - no resistance to malaria parasite (<i>Plasmodium</i>) Data quality - anomaly in first 60 days due to sample size - relevant comment about small sample size for H^SH^S - data obtained from health visitors, not doctors - some health visitors / centres may not respond (therefore not representative) - some health centre data may be estimated / over different timescales / mis-diagnosed - comment about separation of rural and urban areas - relevant comment about length of study Examiner's Comments (c) was a Level of Response item in which candidates were required to describe and explain a set of data as well as comment on the quality of the data. Overall the level of communication was good with the majority producing well-organised and logical responses. Most candidates successfully described the data, made good use of the graph and could make the link between the higher survival rates of those children with sickle cell trait and the associated protection from malaria. However, some candidates described the data without giving any explanations. Weaker explanations were sometimes rather vague and generalised e.g. anaemia leading to less oxygen

Mark Scheme

Question			Answer/Indicative content	Marks	Guidance
					availability or less respiration. There were also significant numbers of candidates who described the resistance to malaria inaccurately as immunity, not comprehending that this is the production of antibodies. Candidates who could give a good scientific explanation of why sickle cell anaemia was life threatening referred to the clumping together of the abnormal haemoglobin and subsequent blockages in capillaries / damage to organs. Likewise, a good scientific explanation for the protection of those with sickle cell trait sometimes referred to peroxide in the cells killing the malarial parasite or referred to an increase in the production of carbon monoxide preventing the development of the disease. A failure by some candidates to read the question carefully resulted in them making no comment on data quality. This did limit their marks. Some good comments on data quality were seen but candidates do need to think carefully about how the data might have been collected and understand the difference between a study of this nature and a controlled experiment in which the independent variable is the only factor which is changed.
			Total	6	
8			D ✓	1	Examiner's Comments This question was more demanding as candidates had to identify which of three statements were correct. Encouragingly more than 50% of candidates chose the correct option on this challenging area of biology.
			Total	1	

Mark Scheme

Question			Answer/Indicative content	Marks	Guidance
9		i	<i>idea that</i> histones are deacetylated / acetyl groups are removed	1	
		ii	<i>inflammatory cell</i> histamine / IgE / cytokine ✓ <i>structural cell</i> mucin / cytokine ✓	2	ALLOW cytokine once only for either type of cell ALLOW a named cytokine
		iii	<i>tissue</i> smooth muscle ✓ <i>location</i> wall of, bronchi / bronchioles ✓	2	
			Total	5	
10		i	0.991 ✓✓	2	If answer incorrect look for $\sqrt{12500} = 0.00894 = q = 1 \text{ mark}$ (for frequency of mutant / low catalase allele) $1 - \sqrt{0.00894} = p$ ALLOW ECF if q calculated incorrectly
		ii	<i>idea that</i> founding population had a high frequency of alleles for low catalase / high proportion of individuals heterozygous or homozygous for low catalase allele ✓	1	IGNORE reference to selective advantage of recessive allele ALLOW ref to 'founder effect'
			Total	3	

Mark Scheme

Question			Answer/Indicative content	Marks	Guidance
11		i	<i>Idea that</i> (disease is) always present in, a population / region;	1	<u>Examiner's Comments</u> Part (i) was synoptic with F222 and relatively few candidates gained a mark here with most confusing the term endemic with pandemic or epidemic.
		ii	1 malaria acts as a selection pressure; 2 mechanism of resistance described; 3 heterozygotes / carriers / sickle cell trait / Hb^sHb^A (are) less likely to get malaria / more likely to survive / have an advantage; 4 sickle cell / advantageous, allele, is passed on in, reproduction / gametes; 5 homozygous 'normal' individuals / Hb^AHb^A, (more likely to) die from malaria; 6 homozygous for sickle cell / Hb^sHb^s, (more likely to) die from (sickle cell) anaemia;		ACCEPT correct use of 'selection' by malaria / Plasmodium ACCEPT idea that parasite / plasmodium / malaria, stays in the body for longer so immunity is enhanced ACCEPT 'those with (one) sickle cell allele' NOTE 'Those having a sickle cell allele have a selective advantage if malaria is present' = 2 marks (1 and 3) ACCEPT 'those with no sickle cell allele' for homozygous normal <u>Examiner's Comments</u> In (ii), few candidates used the idea of malaria acting as a selection pressure and many candidates were not specific enough in describing the heterozygotes or those with the sickle cell trait having the advantage in terms of survival where malaria is present. Some candidates had clearly not covered the learning outcome in F225 and answered in terms of high birth rates and poverty in these regions meaning access to health care was not available.
			Total	5	

Mark Scheme

Question			Answer/Indicative content	Marks	Guidance
12		i	1 <i>idea that</i> (causes) a change in, a DNA, triplet / code / sequence; DNA triplet code / 2 (leading to) a change in, mRNA / codon; 3 (leading to) a change in, tRNA / anticodon (at ribosome); 4 (leading to) a different, amino acid / primary structure / sequence of amino acids; 5 (different amino acids have) different R groups; 6 different bond(ing) in (protein) tertiary structure; 7 <i>idea that</i> (change) leads to (protein) changing its shape / described; 8 consequence for protein action described;	5	ACCEPT idea of 'wrong' or 'incorrect' for 'different' or 'changed' throughout 1. ACCEPT mutation might lead to a stop codon inserted 2. CREDIT idea of a triplet or 3 mRNA bases 6. ACCEPT 'bonding / folding altered in tertiary structure' DO NOT CREDIT 'no protein made' 8 CREDIT examples e.g. (enzyme) no enzyme substrate complex forms, (antibody) variable region not binding to antigen, (protein channel) no hydrophilic R groups lining channel e.g. mutation might lead to a stop codon inserted
		ii	DMD is not a chromosome mutation; (karyotypes) (amniocentesis or CVS) detect, chromosome mutations / described;	1	IGNORE 'DMD is a gene mutation' (as this is given in the stem of the question) CREDIT description e.g. changes in chromosome number or structure.
			Total	6	

Mark Scheme

Question			Answer/Indicative content	Marks	Guidance
13			type 2 / late onset (diabetes); <i>idea that</i> type 2 diabetes results from, changes in metabolism / obesity; <i>idea of</i> genetic influence; (type 2) is not an autoimmune condition;	3 max	2 <i>max</i> for explanation <u>Examiner's Comments</u> Very few candidates understood that poor foetal nutrition would causes changes to metabolism, which is stated in the pre-release material. This question proved challenging and whilst the majority correctly suggested Type 2 diabetes, few went on to explain their choice correctly. Candidates described the reasons for Type 2 diabetes and their explanation referred to the fact that it was late onset. Candidates did not appear to relate this question to any information provided in the pre-release material. A few candidates described Type 2 diabetes as a lack of insulin production.
			Total	3	

Mark Scheme

Question			Answer/Indicative content	Marks	Guidance
14	a	i	0.044 = 2 marks (1)(1)	2	If answer is incorrect look for: 0.21 x 0.21 OR (21/100) x (21/100) OR 4.4% for max 1 mark
		ii	<i>Idea that</i> overall frequency is much lower than expected (1) <i>reason for 0.01 / low frequency</i> more risk of gene mutations occurring (1) <i>idea of</i> base substitution (1) <i>reason for 0.6 / high frequency</i> <i>idea that</i> promotor requires more methylation sites than other regions (1) <i>idea that</i> promoters regulate gene expression (1) <i>idea that</i> RNA polymerase binds to promoter when it is unmethylated (1) methylation allows genes to be switched off when not required (1)	4	DO NOT ALLOW the reverse argument as this is given in the case study
	b	i	<i>Fig. 1.4B (no mark)</i> <i>Explanation:</i> <i>idea that</i> histones still have acetyl groups present (1) <i>idea that</i> chromatin is more open / DNA is more accessible (1) <i>idea that</i> DNA is available for transcription (1) <i>idea that</i> (named) tumour suppressor gene is expressed (1)	2	Maximum of one mark if Fig. 1.4A given as answer ALLOW reverse argument 'acetyl groups have not been removed' ALLOW reference to expression of <i>p53</i> or <i>p16</i> or any correct tumour suppressor gene
		ii	<i>idea that</i> DNMT not available to methylate new strands	1	
			Total	9	

Mark Scheme

Question			Answer/Indicative content	Marks	Guidance
15		i	$q = \sqrt{0.000004} = 0.002$ $p = 1 - 0.002 = 0.998$ $2pq = 2 \times 0.998 \times 0.002 = 0.003992$ % to 1 significant figure = $0.003992 \times 100 = 0.4 \%$	4	Correct answer of 0.4% scores all 4 marks ALLOW ecf for MP 2–4
		ii	Any 2 from: small / decreased, gene pool inbreeding genetic drift population / genetic, bottleneck	2	ALLOW decreased genetic variation IGNORE interbreeding IGNORE references to (increased) homozygous recessive genotypes because this is implied by information provided earlier in the question
			Total	6	
16		i	selection pressure, (is) high altitude / risk of altitude sickness <i>idea of</i> individuals with, mutated gene / allele / Tibetan variant, will survive better at high altitude / have selective advantage <i>idea that</i> allele frequency for Tibetan EPAS1 increases over many generations natural selection	4	
		ii	physiological	1	ALLOW biochemical
		iii	Any 2 from: <i>idea of</i> less oxygen able to be transported in their blood (the potential for) less / reduced / AW, respiration partial pressure / concentration, of oxygen is lower at high altitude	2	
		iv	(erythrocyte cell count using) haemocytometer measures / AW, concentration of (red blood) cells OR flow cytometry measures / AW, volume / morphology / concentration, of (red blood) cells	2	ALLOW numbers of / types of protein in / amount of protein in, (red blood) cells
			Total	9	

Mark Scheme

Question	Answer/Indicative content	Marks	Guidance
17	* Read through the whole answer from start to finish, concentrating on features that make it a stronger or weaker answer using the indicative scientific content as guidance. The indicative scientific content indicates the expected parameters for candidates' answers, but be prepared to recognise and credit unexpected approaches where they show relevance. Using a 'best-fit' approach based on the science content of the answer, first decide which set of level descriptors, Level 1, Level 2 or Level 3, best describes the overall quality of the answer using the guidelines described in the level descriptors in the mark scheme. Once the level is located, award the higher or lower mark. The higher mark should be awarded where the level descriptor has been evidenced and all aspects of the communication statement (in italics) have been met. The lower mark should be awarded where the level descriptor has been evidenced but aspects of the communication statement (in italics) are missing. In summary: • The science content determines the level. • The communication statement determines the mark within a level. Level 3 (5–6 marks) Provides a comprehensive explanation of the speciation process, including the ideas of geographical isolation, selection pressures and adaptations. <i>There is a well-developed line of reasoning which is clear and logically structured and uses scientific terminology at an appropriate level. All the information presented is relevant and forms a</i>	6	Indicative scientific points may include • geographical isolation / allopatric speciation / canyon is physical barrier • (different) selection pressures for each population / in North / South rim areas • example of selection pressure difference • (original) gene pool had allelic variation • different mutations (in separated populations) • alleles / mutations may provide improved adaptation to environment • mutations / alleles passed to offspring • by surviving / selected individuals IGNORE genes passed to offspring • change in allele frequencies / genetic variation between populations • genetic drift • past bottleneck event

Mark Scheme

Question			Answer/Indicative content	Marks	Guidance
			<i>continuous narrative.</i> Level 2 (3–4 marks) Provides an outline of the speciation process, with some ideas about geographical isolation, selection pressures and / or adaptations. <i>There is a line of reasoning presented with some structure and use of appropriate scientific language. The information presented is mostly relevant.</i> Level 1 (1–2 marks) Shows limited understanding of the speciation process, which may include the idea of geographical isolation. <i>There is a logical structure to the answer. The explanation, though basic, is clear.</i> 0 marks No response or no response worthy of credit.		
			Total	6	