1. * The Grand Canyon divides what was once a continuous area of land.

Around 20 million years ago, the Colorado River began to carve into the land to form a deep valley. This valley separated the former continuous land into the North Rim and South Rim.

S. aberti and *S. aberti kaibabensis* form two populations, as shown in Fig. 32.1.

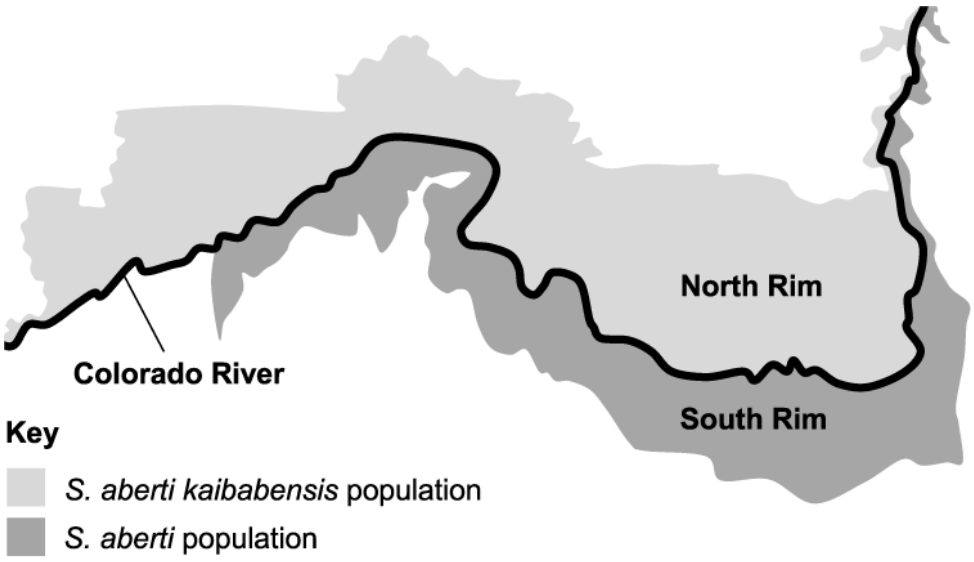

Fig. 32.1

Scientists think that *S. aberti kaibabensis* evolved as a distinct subspecies of *S. aberti*.

Using Fig. 32.1, explain how *S. aberti kaibabensis* could have evolved from *S. aberti*.

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[6]

2. 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]

3. 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.

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

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]**

4. Duchenne's muscular dystrophy (**DMD**) is an inherited disease.

- It is caused by a mutation in a gene that codes for a protein called **dystrophin**.
- Dystrophin is present and functioning in normal muscle cells.
- The malfunctioning or absence of this protein causes muscle weakness that progresses over time.

(i) Explain how a gene mutation can lead to a protein malfunctioning.

[5]

(ii) Suggest why **DMD** cannot be detected by amniocentesis or chorionic villus sampling.

[1]

5. 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]

6. 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.

HBB genotype	Number of children
H^AH^A	392
H^AH^S	374
H^SH^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.

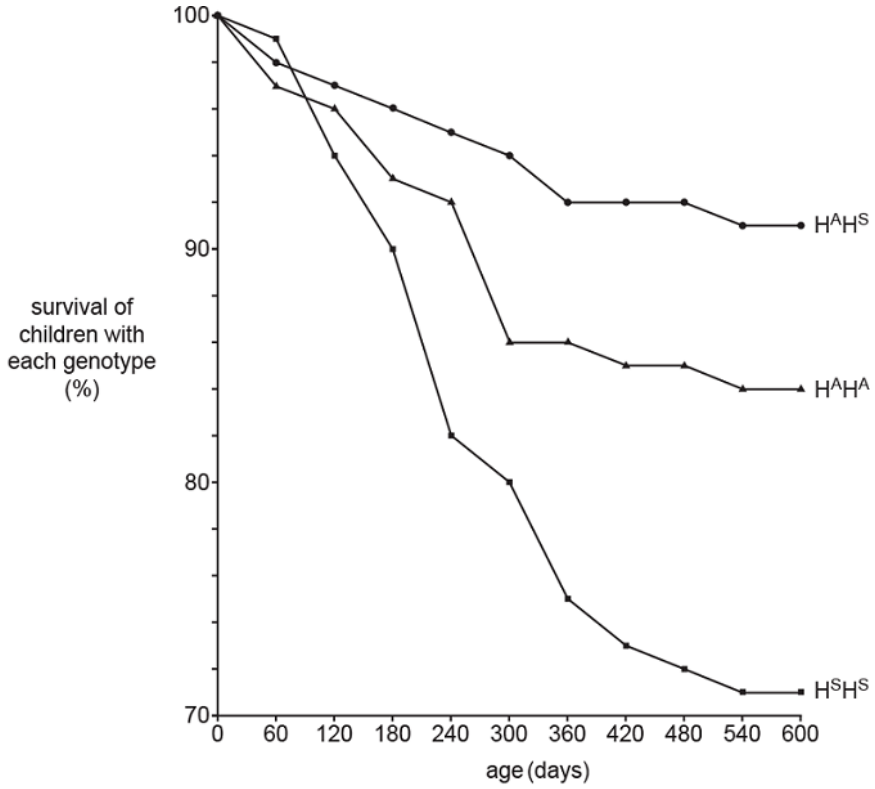

Fig. 31

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]

7. 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]

END OF QUESTION PAPER

Question			Answer/Indicative content	Marks	Guidance
1			* 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. *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*	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

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

Question			Answer/Indicative content	Marks	Guidance
2		i	selection pressure, (is) high altitude / risk of altitude sickness *idea of* individuals with, mutated gene / allele / Tibetan variant, will survive better at high altitude / have selective advantage *idea that* allele frequency for Tibetan EPAS1 increases over many generations natural selection	4	
		ii	physiological	1	ALLOW biochemical
		iii	Any 2 from: *idea of* 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	
3		i	q = √ 0.000004 = 0.002 p = 1 – 0.002 = 0.998 2pq = 2 × 0.998 × 0.002 = 0.003992 % to 1 significant figure = 0.003992 × 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	

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Question			Answer/Indicative content	Marks	Guidance
4		i	1 *idea that* (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 *idea that* (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	

Question			Answer/Indicative content	Marks	Guidance
5		i	*Idea that* (disease is) always present in, a population / region;	1	Examiner's Comments 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 Examiner's Comments 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	

Question			Answer/Indicative content	Marks	Guidance
6			*Summary of instructions to markers:* *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.* *Then, award the higher or lower mark within the level, according to the **Communication Statement** (shown in italics):* o *award the higher mark where the Communication Statement has been met.* o *award the lower mark where aspects of the Communication Statement have been missed.* • ***The science content determines the level.*** • ***The Communication Statement determines the mark within a level.*** **Level 3 (5 – 6 marks)** A detailed description and explanation of the data, comparing the survival of the *HBB* genotypes. There are some valid comments relating to data quality. *There is a well-developed line of reasoning which is clear and logically structured. The information presented is relevant and substantiated.* **Level 2 (3 – 4 marks)** Good description of the data, comparing the survival of the *HBB* genotypes. There is some explanation of survival of at least one genotype OR there is some valid comment on data quality. *There is a line of reasoning presented with some structure. The information presented is in the most-part relevant and supported by some evidence.*	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 (*Plasmodium*) • ref to sickling of erythrocytes in low oxygen tension and destruction of parasite within by phagocytosis

Question	Answer/Indicative content	Marks	Guidance
			• 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
	Level 1 (1 – 2 marks) Good description of the data, comparing the survival of the *HBB* genotypes OR limited explanation of the data. There is no comment on data quality. *There is an attempt at a logical structure with a line of reasoning. The information is in the most part relevant.* 0 marks No response or no response worthy of credit.		H^AH^A genotype • death rate between H^SH^S and H^AH^S ◦ no resistance to malaria parasite (*Plasmodium*) 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

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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	

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Question			Answer/Indicative content	Marks	Guidance
7		i	(drought resistance) involves many, genes / loci ✓ Involves (possible), multiple / variety, alleles (at each locus) ✓	2	ALLOW (drought resistance) is polygenic IGNORE different alleles Examiner's Comments 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 Examiner's Comments 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.

Question			Answer/Indicative content	Marks	Guidance
		iii	place plasmid in Agrobacterium / use gene gun ✓ screen for, transgenic/ recombinant, cells ✓ details of selectable marker ✓	2 max	ALLOW use *Agrobacterium* as vector for 'place plasmid in *Agrobacterium*'. 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	

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