The Roles of Genes in Determining the Phenotype (16.2): Cambridge (CIE) A Level Biology 9700 Exam Questions by

22 questions · 64 marks · about 77 minutes. Answer in the spaces, then check the mark schemes at the end.

Question 1 · Paper 4 · October/November 2022 (42) · Q7(a) · 2 marks

Turn over for Question 7.

7 The determination of sex in domestic turkeys is different from that in humans. The sex chromosomes in turkeys are named **Z** and **W**. Male turkeys are **ZZ** and female turkeys are **ZW**.

The gene for feather colour is located only on the **Z** chromosome.

- The dominant allele codes for bronze feathers.
- The recessive allele codes for brown feathers.

(a) Define the terms dominant and recessive.

dominant .....

.....

recessive .....

.....

[2]

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Question 2 · Paper 4 · October/November 2022 (42) · Q7(b) · 5 marks

(b) Using suitable symbols, construct a genetic diagram to show the results of a cross between a heterozygous bronze male turkey and a brown female turkey.

symbols

parent phenotypes bronze male brown female

parent genotypes

gametes

offspring genotypes

offspring phenotypes

Question 3 · Paper 4 · October/November 2022 (42) · Q7(c) · 3 marks

(c) Explain how you would carry out a test cross to determine the genotype of a bronze male turkey.

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4 The *HFE* gene codes for the HFE protein, which has a role in the regulation of iron absorption by the body. Iron is an essential mineral that can be obtained only from the diet.

A mutation of the *HFE* gene known as C282Y causes hereditary haemochromatosis, which is an autosomal recessive disease. The mutant allele codes for a non-functioning protein. People who are homozygous for the mutant allele produce no functioning HFE protein and this results in an excess of iron being absorbed by the body. The accumulation (build-up) of iron in body organs over many years can cause organ damage.

People that are heterozygous for the *HFE* gene do **not** have hereditary haemochromatosis. They do absorb more iron from their diet than people who do **not** have the mutation, but this does **not** usually have any health effects.

(a) Construct a genetic diagram of a monohybrid cross to show how two parents who do **not** have hereditary haemochromatosis can produce a child with the disease.

Use the following symbols:
H = normal *HFE* allele
h = mutant *HFE* allele.

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3 The tortoise beetle, *Chelymorpha alternans*, is an insect found in Panama that has several different colour patterns. Fig. 3.1 shows a tortoise beetle.

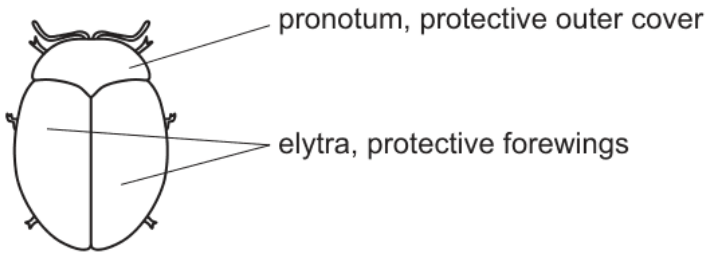


Fig. 3.1

Researchers have identified a gene, ***L***, that controls colour pattern in the pronotum and elytra. Gene ***L*** has four different alleles: L^V, L^T, L^R and L^r.

Table 3.1 shows five different colour pattern phenotypes of tortoise beetles and their genotypes.

Table 3.1

phenotype		genotypes
rufipennis	 black; red	$L^R\ L^R$ $L^R\ L^V$ $L^R\ L^r$
darien f. militaris-a (**dfm-a**)	 black and red striped	$L^T\ L^T$ $L^T\ L^V$ $L^T\ L^r$
darien f. militaris-b (**dfm-b**)	 black; black and red striped	$L^T\ L^R$
veraguensis	 red	$L^V\ L^V$ $L^V\ L^r$
metallic	 metallic and red striped	$L^r\ L^r$

(a) Explain why the inheritance of colour pattern in tortoise beetles can be described as involving multiple alleles.

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Question 6 · Paper 4 · May/June 2024 (42) · Q3(b) · 4 marks

(b) A tortoise beetle with **dfm-b** phenotype was crossed with another tortoise beetle with **dfm-b** phenotype.

Construct a genetic diagram to show the results of this cross, including the ratio of offspring phenotypes.

parental phenotypes: **dfm-b** × **dfm-b**

parental genotypes:

gametes:

offspring genotypes:

offspring phenotypes:

ratio of offspring phenotypes:

Question 7 · Paper 4 · May/June 2024 (42) · Q3(c) · 2 marks

(c) Colour pattern phenotype involves alleles that show codominance. There is also an order of dominance of alleles (dominance hierarchy).

Use the information in Table 3.1 to:

- identify the codominant alleles
- list the dominance hierarchy with alleles from the most dominant to the least dominant.

codominant alleles .....

dominance hierarchy .....

[2]

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Question 8 · Paper 4 · May/June 2024 (42) · Q3(d)(i) · 2 marks

(d) Researchers carried out two crosses.

Cross 1: female veraguensis tortoise beetles were crossed with male metallic tortoise beetles.

The results are shown in Table 3.2.

Table 3.2

number of observed offspring phenotypes				ratio of observed colour pattern phenotypes
male veraguensis	female veraguensis	male metallic	female metallic	
139	153	136	140	1.06 : 1

Cross 2: female veraguensis tortoise beetles were crossed with male veraguensis tortoise beetles.

The results are shown in Table 3.3.

Table 3.3

number of observed offspring phenotypes				ratio of observed colour pattern phenotypes
male veraguensis	female veraguensis	male metallic	female metallic	
693	592	237	213	2.9 : 1

(i) Using Table 3.1, deduce the genotypes of **each** of the parental beetles used in cross 1 and cross 2.

cross 1 .....

cross 2 .....

[2]

Question 9 · Paper 4 · May/June 2024 (42) · Q3(d)(ii) · 1 mark

(ii) An assumption was made that female tortoise beetles have XX chromosomes and males have XY chromosomes. Gene ***L*** is **not** located on the X chromosome. It was concluded that colour pattern phenotype followed autosomal inheritance.

Explain how the evidence in Table 3.3 supports this conclusion.

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Question 10 · Paper 4 · May/June 2025 (42) · Q3(a)(i) · 5 marks

3 Rice, *Oryza sativa*, is an important grain crop. A rice grain is a seed and can have a structure known as an awn, which projects from the tip of the grain.

Fig. 3.1 shows a rice grain with an awn present and a rice grain with no awn present.

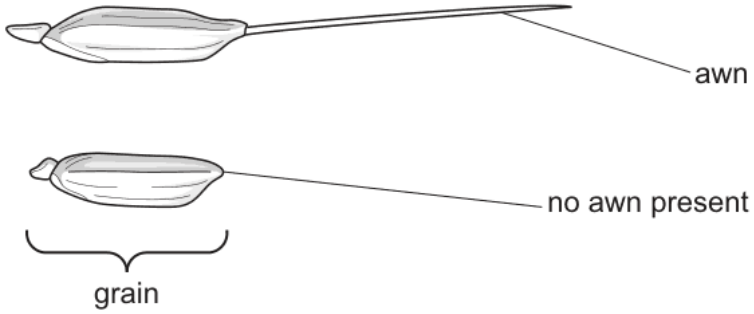


Fig. 3.1

(a) (i) The development of awns is controlled by two genes: gene **A/a** and gene **B/b**. The genes are present on different autosomes. The presence of either dominant allele **A** or dominant allele **B** results in rice plants producing grains with awns.

A cross was carried out between a rice plant that is homozygous dominant for the two genes (double homozygous dominant) and a rice plant that is homozygous recessive for the two genes (double homozygous recessive). All F1 offspring plants produced grains with awns.

Construct a genetic diagram, including a Punnett square, to show the cross that produces the F2 generation, including phenotypes.

State the ratio of the offspring phenotypes produced.

Question 11 · Paper 4 · May/June 2025 (42) · Q3(a)(ii) · 1 mark

(ii) Deduce the type of inheritance shown by the ratio of the offspring phenotypes stated in **3(a)(i)**.

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5 The fruit fly, *Drosophila melanogaster*, has autosomal genes for body colour and wing shape.

Gene **B/b** is involved in the production of body colour:
- **B** = dominant allele for brown body colour
- **b** = recessive allele for black body colour.

Gene **D/d** is involved in wing shape:
- **D** = dominant allele for straight wing
- **d** = recessive allele for curved wing.

A dihybrid test cross was carried out between flies heterozygous for body colour and for wing shape and flies homozygous recessive for body colour and for wing shape.

(a) Table 5.1 shows the number of offspring of each phenotype obtained in the test cross.

Table 5.1

phenotype	observed number	expected number
brown body colour, straight wings	2843	
brown body colour, curved wings	855	
black body colour, straight wings	842	
black body colour, curved wings	2768	

Use Table 5.1 to calculate the expected number of each phenotype if the two genes are on **different** autosomes. Write your answers in the table. [1]

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(b) A chi-squared (χ^2) test was carried out to compare the observed results with the results that would be expected from a dihybrid cross involving genes on different autosomes.

The value of χ^2 = 2097.836.

Table 5.2 shows the critical values for the χ^2 distribution.

Table 5.2

degrees of freedom	*p* value		
	0.05	**0.01**	**0.001**
1	3.841	6.635	10.828
2	5.991	9.210	13.816
3	7.815	11.345	16.266
4	9.488	13.277	18.467

Explain how the value of χ^2 and Table 5.2 can be used to assess the significance of the difference between the observed results and the expected numbers in Table 5.1.

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Question 14 · Paper 4 · October/November 2023 (42) · Q5(c) · 4 marks

(c) Suggest explanations for the **observed** results in Table 5.1.

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(b) Wing pattern in the butterfly species *Heliconius melpomene* is controlled by genes on autosomal chromosomes.

The gene for banding pattern on the upper wing has two alleles:

- a dominant allele coding for a full band
- a recessive allele coding for a broken band.

The gene for ray pattern on the lower wing has two alleles:

- a dominant allele coding for rays
- a recessive allele coding for no rays.

Scientists crossed a butterfly that was homozygous dominant for both genes with a butterfly that was homozygous recessive for both genes. The scientists wanted to check whether the phenotypic ratio for offspring in the F2 generation agreed with the expected phenotypic ratio of 9 : 3 : 3 : 1.

The results of this genetic cross are shown in Fig. 4.2.

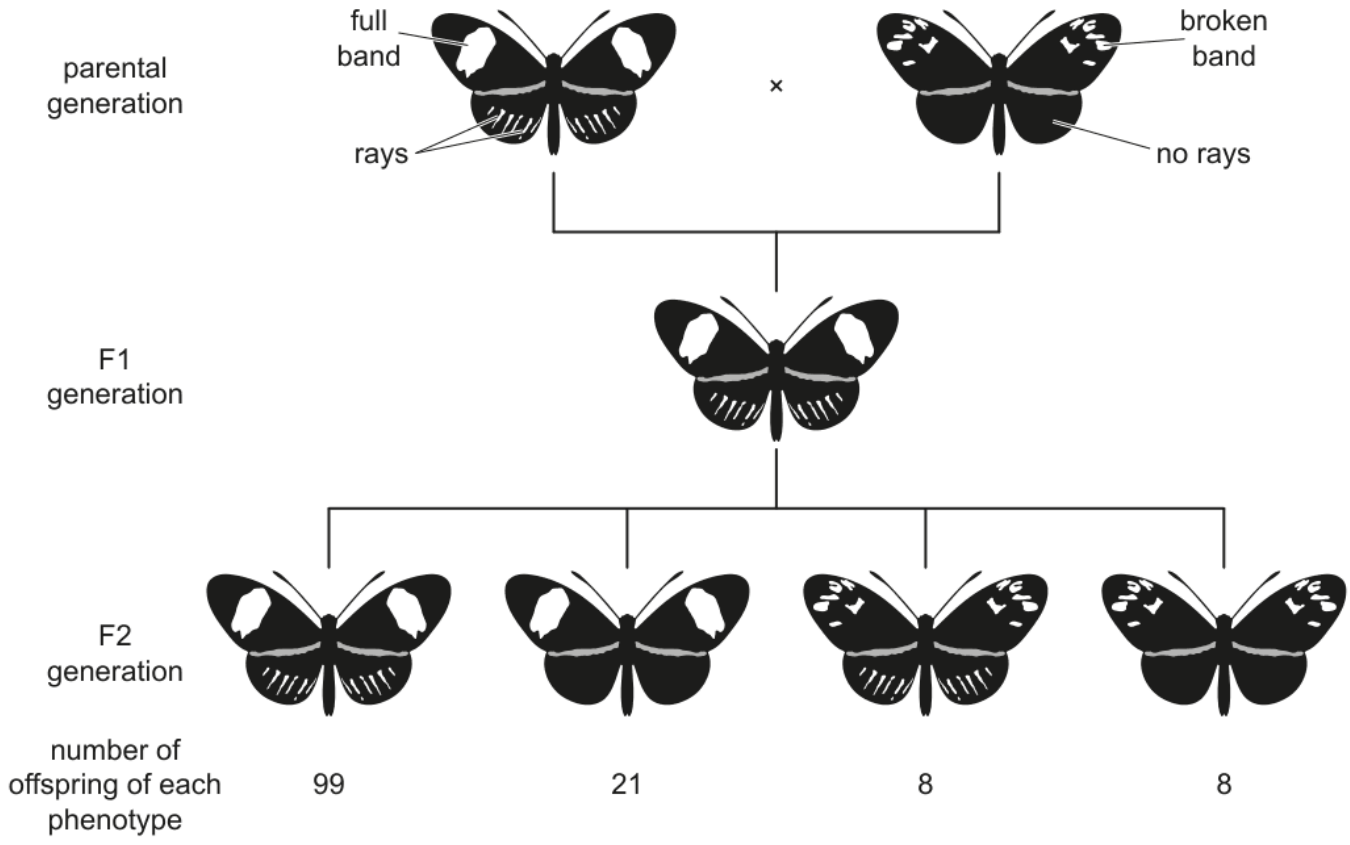


Fig. 4.2

(i) Explain the term F1 generation.

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(ii) The scientists used the chi-squared (χ^2) test to compare their data to the expected phenotypic ratio of 9 : 3 : 3 : 1. The formula for the chi-squared test is shown in Fig. 4.3.

$$\chi^2 = \sum \frac{(O-E)^2}{E}$$

key to symbols

O = observed value

E = expected value

Fig. 4.3

Complete Table 4.1 and use the chi-squared formula in Fig. 4.3 to calculate the χ^2 value for these data.

Table 4.1

phenotype	expected ratio	O	E	$O-E$	$(O-E)^2$	$\frac{(O-E)^2}{E}$
full band and rays	9	99	76.5	22.5	506.25	6.62
full band and no rays	3	21	25.5	−4.5	20.25	0.79
broken band and rays	3	8	.....	.....	.....	.....
broken band and no rays	1	8	8.5	−0.5	0.25	0.03

χ^2 = ...

[3]

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Question 17 · Paper 4 · February/March 2022 (42) · Q4(b)(iii) · 3 marks

(iii) The critical value at the 0.05 probability level and three degrees of freedom is 7.81.

Using the result of your calculation in **(b)(ii)**, explain whether the results of the study agree with the expected ratio of phenotypes for the F2 generation.

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Question 18 · Paper 4 · February/March 2022 (42) · Q4(b)(iv) · 2 marks

(iv) Suggest **two** reasons why phenotypic ratios in the F2 generation do **not** always match the expected ratios.

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Question 19 · Paper 4 · February/March 2025 (42) · Q10(b) · 3 marks

(b) Some of the neurotransmitters in the brain are produced through a series of reactions (reaction pathway) from a chemical called DOPA.

DOPA is also involved in other reaction pathways. For example, in the skin and eyes, DOPA is part of a different reaction pathway that depends on the *TYR* gene.

Describe **and** explain the phenotypic consequences for the skin and eyes of a person who is homozygous for a mutated, non-functional allele of the *TYR* gene.

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Question 20 · Paper 4 · February/March 2022 (42) · Q3(a) · 5 marks

3 The red blood cells of people with sickle cell anaemia have reduced oxygen-carrying capacity.

(a) Explain the relationship between the gene mutation that causes sickle cell anaemia and the reduced oxygen-carrying capacity of red blood cells.

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Question 21 · Paper 4 · May/June 2024 (42) · Q4(a) · 5 marks

4 Genes have a role in determining the overall phenotype of an organism.

(a) Using haemophilia as an example, explain the relationship between a gene, a protein and a phenotype.

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Question 22 · Paper 4 · May/June 2024 (42) · Q4(b) · 3 marks

(b) In the pea plant, *Pisum sativum*, the genotype ***lele*** results in plants with short stems (dwarf plants).

Explain how the ***lele*** genotype and its gene product results in dwarf plants.

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Mark schemes

Question 1 · Paper 4 · October/November 2022 (42) · Q7(a) · 2 marks

7(a)	*dominant* – allele always has effect on phenotype / allele expressed in homozygote and heterozygote / allele always expressed ; *recessive* – allele only has effect on phenotype if dominant allele absent / only expressed in homozygote ;	2

Question 2 · Paper 4 · October/November 2022 (42) · Q7(b) · 5 marks

7(b)	*symbols* Z^B = allele for bronze feathers Z^b = allele for brown feathers ; *parents phenotypes* *bronze male* x *brown female* *parents genotypes* Z^BZ^b x Z^bW ; *gametes* Z^B Z^b Z^b W ; *offspring genotypes* Z^BZ^b Z^BW Z^bZ^b Z^bW ; *offspring phenotypes* bronze male, bronze female, brown male, brown female; *ecf for putting allele on **W*** *ecf for incorrect symbols with no superscript* *ecf for different letters for alleles* *no sex chromosomes = no marks*	5

Question 3 · Paper 4 · October/November 2022 (42) · Q7(c) · 3 marks

7(c)	1 cross with brown female ; 2 if **all** offspring bronze then male is, homozygous / Z^BZ^B ; 3 if (some offspring are bronze and) some are brown then male is, heterozygous / Z^BZ^b ;	3

Question 4 · Paper 4 · February/March 2025 (42) · Q4(a) · 3 marks

4(a)	*parental gametes* H h × H h ; **accept** from Punnett square *offspring genotypes* HH Hh Hh hh ; **accept** from Punnett square *offspring phenotypes* no disease, disease / haemochromatosis ;	3

Question 5 · Paper 4 · May/June 2024 (42) · Q3(a) · 1 mark

3(a)	(gene ***L*** has) four alleles / L^V, L^T, L^R and L^r ; **A** a gene which has more than two alleles	1

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Question 6 · Paper 4 · May/June 2024 (42) · Q3(b) · 4 marks

3(b)	parental genotypes: $L^T L^R$ x $L^T L^R$ gametes: (L^T) (L^R) (L^T) (L^R) ; offspring genotypes: $L^T L^T$ $L^T L^R$ $(L^T L^R)$ $L^R L^R$; offspring phenotypes: dfm-a dfm-b (dfm-b) rufipennis ; linked to genotypes ratio of offspring phenotypes: 1 : 2 : 1 ; *accept correct phenotype descriptions*	4

Question 7 · Paper 4 · May/June 2024 (42) · Q3(c) · 2 marks

3(c)	L^T and L^R ; L^T and $L^R > L^V > L^r$;	2

Question 8 · Paper 4 · May/June 2024 (42) · Q3(d)(i) · 2 marks

3(d)(i)	(female) $L^V L^r$ and (male) $L^r L^r$; $L^V L^r$ (and $L^V L^r$) ;	2

Question 9 · Paper 4 · May/June 2024 (42) · Q3(d)(ii) · 1 mark

3(d)(ii)	there are similar numbers of male and female offspring produced (for each phenotype) **or** female metallic offspring are produced **or** if linked to X chromosome no female metallic offspring are produced ;	1

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Question 10 · Paper 4 · May/June 2025 (42) · Q3(a)(i) · 5 marks

3(a)(i)

F1 genotypes: AaBb x AaBb ; **5**

gametes: AB Ab aB ab (AB Ab aB ab) ;

F2 genotypes:

	AB	**Ab**	**aB**	**ab**	
AB	AABB awn	AABb awn	AaBB awn	AaBb Awn	
Ab	AABb awn	AAbb awn	AaBb awn	Aabb awn	
aB	AaBB awn	AaBb awn	aaBB awn	aaBb awn	
ab	AaBb awn	Aabb awn	aaBb awn	aabb no awn	;

F2 phenotypes linked to genotypes (see Punnet Square) ;

ratio of F2 phenotypes: 15:1
awn: no awn ;

Question 11 · Paper 4 · May/June 2025 (42) · Q3(a)(ii) · 1 mark

3(a)(ii)

autosomal dominant / epistasis ; **1**
I dihybrid

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Question 12 · Paper 4 · October/November 2023 (42) · Q5(a) · 1 mark

5(a) all correct (1827) ; **1**

phenotype	observed number	expected number
brown body colour, straight wings	2843	1827
brown body colour, curved wings	855	1827
black body colour, straight wings	842	1827
black body colour, curved wins	2768	1827

Question 13 · Paper 4 · October/November 2023 (42) · Q5(b) · 4 marks

5(b) *any **four** from:* **4**

1 *ref. to* 3 degrees of freedom ;
2 the, χ^2 value / 2098, is greater than 7.815 (at p=0.05) ;
3 there is a significant difference ;
4 reject the null hypothesis ;
5 any difference is not due to chance / less than 5% (probability) that difference is due to chance ;
6 the, χ^2 value / 2098, is greater than, 11.345 at p=0.01 / 16.266 at p=0.001
or
less than, 1% / 0.1% (probability), that difference is due to chance ;

Question 14 · Paper 4 · October/November 2023 (42) · Q5(c) · 4 marks

5(c) *any **four** from:* **4**

1 greater numbers of parental phenotypes / fewer numbers of recombinants ;
2 genes are linked / autosomal linkage / genes are on same chromosome ;
3 alleles inherited together ;
4 **B** inherited with **D** and **b** inherited with **d**
or
B and **D** in the same gamete and **b** and **d** in the same gamete ;
5 no independent assortment ;
6 crossing over (produces recombinants) ;
7 during, meiosis / prophase I / gamete formation ;
8 AVP ; e.g. idea that genes are far enough apart on chromosome for recombination to occur

Question 15 · Paper 4 · February/March 2022 (42) · Q4(b)(i) · 2 marks

4(b)(i) *any **two** from:* **2**

1 (first) generation of offspring (from parental cross) ;
2 (parents are) homozygous / true bred / pure bred / pure breeding ;
3 F1 generation will be heterozygous ;

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Question 16 · Paper 4 · February/March 2022 (42) · Q4(b)(ii) · 3 marks

4(b)(ii) **3**

phenotype	expected ratio	O	E	$O-E$	$(O-E)^2$	$\frac{(O-E)^2}{E}$
full band and rays	9	99	76.5	22.5	506.25	6.62
full band and no rays	3	21	25.5	−4.5	20.25	0.79
broken band and rays	3	8	25.5	−17.5	306.25	12.01
broken band and no rays	1	8	8.5	−0.5	0.25	0.03

1 mark for E and $O-E$ correct ;

1 mark for $(O-E)^2$ and $\frac{(O-E)^2}{E}$ correct ;

χ^2 = 19.45 ;

ecf for 1 mistake in one box

Question 17 · Paper 4 · February/March 2022 (42) · Q4(b)(iii) · 3 marks

4(b)(iii) **3**

*any **three** from:*

1 19.45 / chi-squared value, is greater than, 7.81 / critical value (at p = 0.05) ;

2 results are / observed ratio is, significantly different to expected ratio ;

3 so differences are not due to chance ;

4 due to, increase in full band phenotypes / decrease in broken band phenotypes ;

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Question 18 · Paper 4 · February/March 2022 (42) · Q4(b)(iv) · 2 marks

4(b)(iv)	*any **two** from:* 1 epistasis ; 2 linkage ; 3 lethal allele ; 4 chance deviations ; 5 environmental effects ; 6 mutations ; 7 small sample size ;	**2**

Question 19 · Paper 4 · February/March 2025 (42) · Q10(b) · 3 marks

10(b)	*any **three** from:* 1 albino / albinism **or** described ; e.g. no pigment or melanin in skin / pale skin / white hair 2 poor vision **or** described ; e.g. red or pink eyes / irises, eyes sensitive to light / jerky eye movements 3 tyrosinase, non-functioning / not produced ; 4 tyrosine not converted to, DOPA / dopaquinone ; 5 no melanin produced ;	**3**

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Question 20 · Paper 4 · February/March 2022 (42) · Q3(a) · 5 marks

3(a)	*any **five** from:* *max **four** from mp1–mp6:* 1 (sickle cell anaemia is) homozygous recessive ; 2 ref. to beta / β, globin / polypeptide **or** *HBB* (allele / gene) ; 3 base / nucleotide, substitution **or** mis-sense mutation **or** change in, triplet / codon **or** GAG to GTG / A to T ; 4 causes, change in amino acid (sequence) / glutamic acid to valine ; 5 valine is non-polar ; 6 causes change in, 3D shape / tertiary structure / quaternary structure / globular structure (of haemoglobin) ; 7 haemoglobin has lower affinity for oxygen ; 8 (in red blood cells) haemoglobin, less soluble / sticks together / forms fibres ; 9 (red blood) cells become sickle shaped ;	**5**

Question 21 · Paper 4 · May/June 2024 (42) · Q4(a) · 5 marks

4(a)	*any **five** from:* 1 a gene codes for a protein which determines the phenotype ; 2 *F8* gene ; 3 located on X chromosome / sex-linked ; 4 *ref. to* recessive allele ; 5 non-functioning factor VIII (protein) **or** less factor VIII (protein) **or** no factor VIII (protein) ; 6 (so) blood does not clot quickly enough / excessive bleeding occurs (after an injury) ; 7 link the phenotype to the genotype ; e.g. $X^F X^F$ = normal female $X^F X^f$ = normal female $X^f X^f$ = affected female $X^F Y$ = normal male $X^f Y$ = affected male 8 AVP ; e.g. prevents activation of thrombin / fibrinogen not converted to fibrin / *ref. to* mostly males affected / different mutations cause a range of severity of haemophilia	**5**

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Question 22 · Paper 4 · May/June 2024 (42) · Q4(b) · 3 marks

4(b)	*any* ***three*** *from:* 1 *lele* is, homozygous recessive / two recessive alleles **;** 2 *le* (allele) / *lele*, codes for a non-functional <u>enzyme</u> **;** 3 no, active gibberellin / GA_1 **or** *ref. to* gibberellin inactive **;** 4 no / less, cell elongation **or** no / less, stem elongation / increase in internodal length **;** 5 AVP **;** e.g. *ref. to* GA 3β-hydroxylase / GA3-oxidase / last enzyme in the pathway that produces, active gibberellin / GA_1 *le* allele codes for alanine instead of threonine (in the enzyme)	**3**

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