

AQA A-LEVEL • 7402

BIOLOGY



EXAM QUESTION PACK

3.7.1 INHERITANCE AND GENETIC CROSSES

SCORE

/ 429

PERCENTAGE

%

STEMUP

These questions belong to AQA, not to us.

The examination questions, mark schemes and examiner reports in this pack were written and published by AQA. They are past assessment materials, reproduced here so that students can revise from them.

What we do not own

STEMUP is not affiliated with AQA and does not own, and makes no claim to own, any of the question, mark scheme or examiner report material in this pack. AQA has not endorsed, approved or been involved in the production of it. AQA and the qualification titles used here belong to their respective owners and appear for identification only.

Where to find the originals

AQA publishes its past papers, mark schemes and examiner reports free of charge. Always check the current specification at aqa.org.uk before you revise, because content and wording can change between series.

If you hold the rights to any material in this pack and would like it amended or removed, contact admin@stemup.co.uk and we will act on it.

Q1.

- (a) In genetic crosses, the observed phenotypic ratios obtained in the offspring are often **not** the same as the expected ratios.

Suggest **two** reasons why.

1. _____

2. _____

(2)

In tomato plants, the genes for height and for the type of leaf are on the same homologous pair of chromosomes. The allele **T**, for a tall plant, is dominant to the allele **t**, for a dwarf plant. The allele **M**, for normal leaves, is dominant to the allele **m**, for mottled leaves.

A biologist carried out crosses between parent plants heterozygous for both genes and examined the offspring produced. The position of the two alleles for both genes was the same in each parent plant as shown in the diagram. The phenotypes and number of offspring produced are shown in **Table 1**.

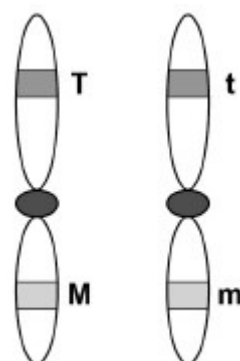


Table 1

Phenotype of offspring	Number of offspring
Tall plants and normal leaves	1860
Tall plants and mottled leaves	68
Dwarf plants and normal leaves	57
Dwarf plants and mottled leaves	580

- (b) What would be the genotype of the offspring with dwarf plants and mottled leaves?

(1)

- (c) Use the information provided to explain the results in **Table 1**.

[illegible]

- (d) Complete **Table 2** to show the expected ratio of phenotypes if the same cross had been carried out but the genes for height of plant and for the type of leaf were on different homologous pairs of chromosomes.

Table 2

Phenotype of offspring	Ratio of offspring

Lactose is the main sugar in milk and is hydrolysed by the enzyme lactase. Lactase is essential to newborn mammals as milk is their only source of food. Most mammals stop producing lactase when they start feeding on other food sources. Humans are an exception to this because some continue to produce lactase as adults. The ability to continue producing lactase is known as lactase persistence (LP) and is controlled by a dominant allele. A number of hypotheses based on different selection pressures have been put forward to explain LP in humans.

Describe how farming cattle as a source of milk could have led to an increase in LP.

(4)

- (b) Use the information provided to explain why the number of people showing LP would **rapidly** increase once selection for this condition had been established.

(2)

- (c) Lactase persistence is caused by a mutation in DNA. This mutation does not occur in the gene coding for lactase.

Suggest and explain how this mutation causes LP.

(2)

(Total 8 marks)

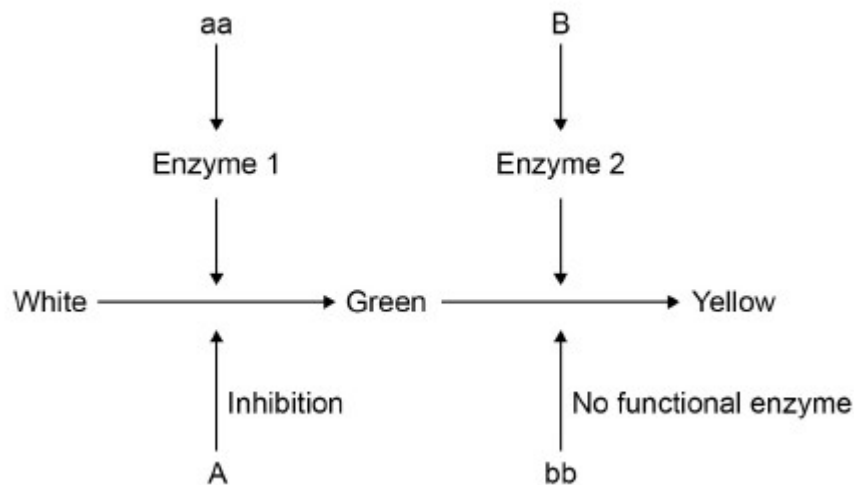
Q3.

- (a) What is meant by the term phenotype?

(2)

- (b) The inheritance of fruit colour in summer squash plants is controlled by two genes, **A** and **B**. Each gene has two alleles.

The diagram shows the interaction of these two genes in controlling fruit colour in summer squash plants.



Name the type of gene interaction shown in the diagram above.

(1)

- (c) What fruit colour would you expect the following genotypes to have?

AAbb _____

aaBB _____

(1)

- (d) Genes **A** and **B** are not linked.

Complete the genetic diagram to show all the possible genotypes and the ratio of phenotypes expected in the offspring of this cross.

Genotypes of parents **aabb** × **AaBb**

Genotypes of offspring _____

Phenotypes of offspring _____

Ratio of phenotypes _____

(3)

- (e) A population of summer squash plants produced only green and yellow fruit. The percentage of plants producing yellow fruit in this population was 36%.

Use the Hardy–Weinberg equation to calculate the percentage of plants that were heterozygous for gene **B**.

Answer = _____ %

(2)

(Total 9 marks)

Q4.

A student investigated the monohybrid inheritance of eye shape in fruit flies. Two fruit flies with bar (narrow) eyes were crossed. Of the offspring, 1538 had bar eyes and 462 had round (normal) eyes.

- (a) Using suitable symbols, give the genotypes of the parents.

Explain your answer.

Genotypes _____

Explanation _____

(2)

- (b) The ratio of bar-eyed flies and round-eyed flies in the student's results were not the same as the ratio she had expected.

What ratio of bar-eyed to round-eyed flies was the student expecting?

(1)

- (c) Suggest **two** reasons why observed ratios are often **not** the same as expected ratios.

(2)

- (d) The student wished to test her results with the ones she had expected.
Which statistical test should she use?

(1)

- (e) This fruit fly has another characteristic controlled by a pair of codominant alleles, W^N and W^V .

What is meant by **codominant** alleles?

(1)

- (f) There were 850 fruit flies in one population. In this population, 510 fruit flies had the genotype $W^N W^N$, 255 had the genotype $W^N W^V$ and 85 had the genotype $W^V W^V$.

Calculate the **actual** frequency of the allele W^V . **Do not** use the Hardy-Weinberg equation in your calculation.

Answer = _____

(1)

- (g) In another population of 950 fruit flies, the frequency of the W^V allele was 0.2.

Use the Hardy-Weinberg equation to calculate the number of insects that would be **expected** to have the genotype $W^N W^V$.

Answer = _____

(2)

(Total 10 marks)

Q5.

- (a) In fruit flies, the genes for body colour and wing length are linked. Explain what this means.

(1)

A scientist investigated linkage between the genes for body colour and wing length. He carried out crosses between fruit flies with grey bodies and long wings and fruit flies with black bodies and short wings.

Figure 1 shows his crosses and the results.

- **G** represents the dominant allele for grey body and **g** represents the recessive allele for black body.
- **N** represents the dominant allele for long wings and **n** represents the recessive allele for short wings.

Figure 1

<i>Phenotype of parents</i>	grey body, long wings	×	black body, short wings
<i>Genotype of parents</i>	GGNN		ggnn
<i>Genotype of offspring</i>	GgNn		
<i>Phenotype of offspring</i>	all grey body, long wings		

These offspring were crossed with flies homozygous for black body and short wings.

The scientist's results are shown in **Figure 2**.

Figure 2

	GgNn	crossed with	ggnn	
	Grey body, long wings	Black body, short wings	Grey body, short wings	Black body, long wings
Number of offspring	975	963	186	194

- (b) Use your knowledge of gene linkage to explain these results.

(4)

- (c) If these genes were **not** linked, what ratio of phenotypes would the scientist have expected to obtain in the offspring?

(1)

- (d) Which statistical test could the scientist use to determine whether his observed results were significantly different from the expected results?

Give the reason for your choice of statistical test.

(2)

(Total 8 marks)

Q6.

Read the following passage carefully.

A large and growing number of disorders are now known to be due to types of mitochondrial disease (MD). MD often affects skeletal muscles, causing muscle weakness.

We get our mitochondria from our mothers, via the fertilised egg cell. Fathers do not pass on mitochondria via their sperm. Some mitochondrial diseases are caused by mutations of mitochondrial genes inside the mitochondria. Most mitochondrial diseases are caused by mutations of genes in the cell nucleus that are involved in the functioning of mitochondria. These mutations of nuclear DNA produce

5

recessive alleles.

One form of mitochondrial disease is caused by a mutation of a mitochondrial gene that codes for a tRNA. The mutation involves substitution of guanine for adenine in the DNA base sequence. This changes the anticodon on the tRNA. This results in the formation of a non-functional protein in the mitochondrion.

There are a number of ways to try to diagnose whether someone has a mitochondrial disease. One test involves measuring the concentration of lactate in a person's blood after exercise. In someone with MD, the concentration is usually much higher than normal. If the lactate test suggests MD, a small amount of DNA can be extracted from mitochondria and DNA sequencing used to try to find a mutation.

Use information in the passage and your own knowledge to answer the following questions.

- (a) Mitochondrial disease (MD) often causes muscle weakness (lines 1–3). Use your knowledge of respiration and muscle contraction to suggest explanations for this effect of MD.

(3)

Two couples, couple **A** and couple **B**, had one or more children affected by a mitochondrial disease. The type of mitochondrial disease was different for each couple.

None of the parents showed signs or symptoms of MD.

- Couple **A** had four children who were all affected by an MD.
- Couple **B** had four children and only one was affected by an MD.

- (b) Use the information in lines 5–9 and your knowledge of inheritance to suggest why:

- all of couple **A**'s children had an MD
- only one of couple **B**'s children had an MD.

Couple **A** _____

Couple B _____

(4)

- (c) Suggest how the change in the anticodon of a tRNA leads to MD (lines 10–13).

(3)

- (d) If someone has MD, the concentration of lactate in their blood after exercise is usually much higher than normal (lines 15–17). Suggest why.

(3)

- (e) A small amount of DNA can be extracted from mitochondria and DNA sequencing used to try to find a mutation (lines 18–19).

From this sample:

- how would enough DNA be obtained for sequencing?
- how would sequencing allow the identification of a mutation?

(2)

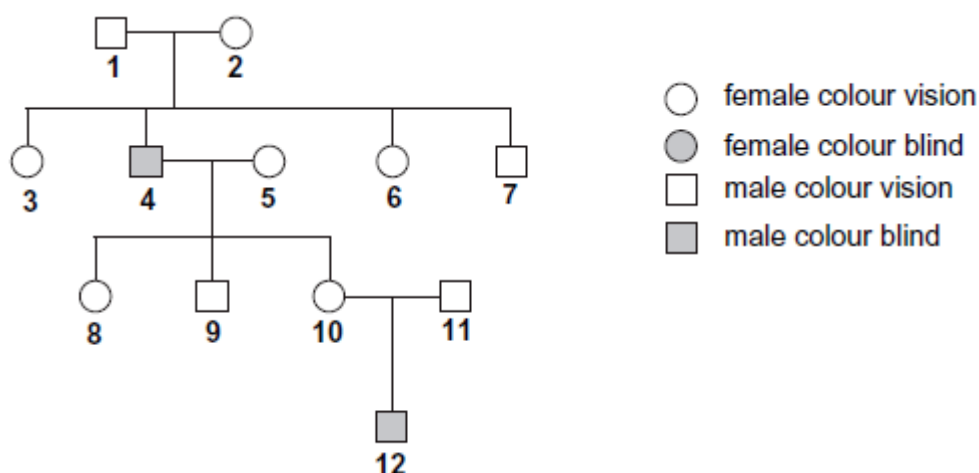
Q7.

- (a) Explain what is meant by the term phenotype.

(2)

- (b) One type of colour blindness is controlled by a gene carried on the X chromosome. The allele for this type of colour blindness, **b**, is recessive to the allele for colour vision, **B**.

The diagram shows the phenotypes in a family tree for this sex-linked condition.



- (i) Explain **one** piece of evidence from the diagram which shows that colour blindness is recessive.

(2)

- (ii) Give the genotype of individual 8.

(1)

- (c) (i) The allele for tongue-rolling, **T**, is dominant to the allele for non-tongue rolling, **t**.

The gene controlling tongue-rolling is **not** sex-linked. Individuals **10** and **11** are

both heterozygous for tongue-rolling.

What is the probability that individuals **10** and **11** will produce a male child who is colour blind and a non-tongue roller?

Answer = _____

(2)

- (ii) In a population, the frequency of the allele for tongue-rolling, **T**, is 0.4.

Use the Hardy-Weinberg equation to calculate the percentage of people in this population that are heterozygous for tongue-rolling.

Answer = _____ %

(2)

(Total 9 marks)

Q8.

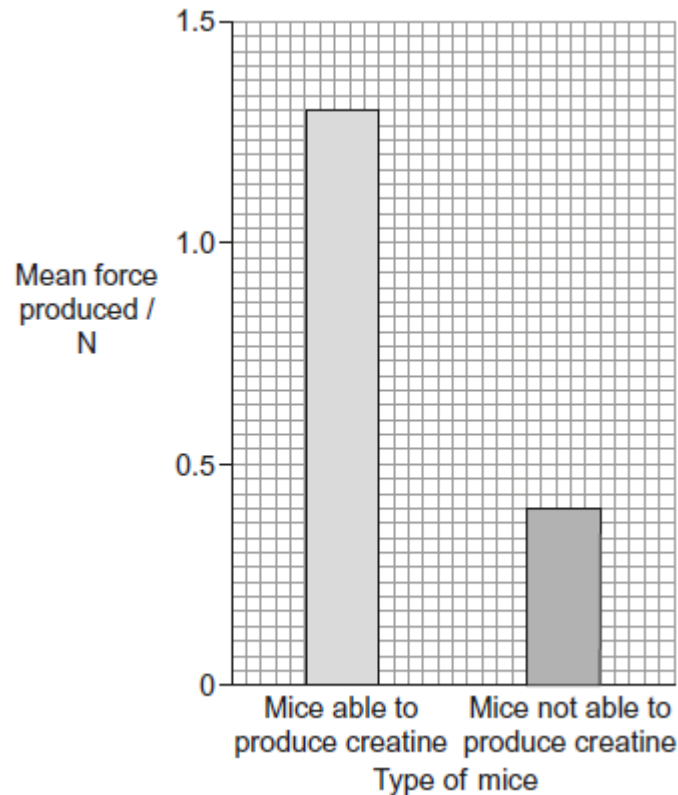
- (a) What is the role of ATP in myofibril contraction?

(2)

- (b) Scientists investigated the effect of not being able to produce creatine on the force produced by muscle. They used mice with a mutation that made them not able to produce creatine.

The force produced when these mice gripped with their paws was compared with the force produced by normal mice that were able to produce creatine.

The graph shows the scientists' results.



- (i) What was the percentage fall in the mean force produced by mice not able to produce creatine, compared with the normal mice? Show your working.

Answer _____ %

(2)

- (ii) Suggest an explanation for these results.

(2)

- (c) The mice that were not able to produce creatine were homozygous for a recessive allele of a gene. Mice that are heterozygous for this allele are able to produce forces similar to those of normal mice that are homozygous for the dominant allele of the same gene.

Explain why the heterozygous mice can produce forces similar to those of normal mice.

(2)

(Total 8 marks)

Q9.

In cats, males are XY and females are XX. A gene on the X chromosome controls fur colour in cats. The allele **G** codes for ginger fur and the allele **B** codes for black fur. These alleles are codominant. Heterozygous females have ginger and black patches of fur and their phenotype is described as tortoiseshell.

- (a) Explain what is meant by **codominant** alleles.

(1)

- (b) Male cats with a tortoiseshell phenotype do **not** usually occur. Explain why.

(1)

- (c) A tortoiseshell female was crossed with a black male. Use a genetic diagram to show all the possible genotypes and the ratio of phenotypes expected in the offspring of this cross.

Use X^G to indicate the allele **G** on an X chromosome.

Use X^B to indicate the allele **B** on an X chromosome.

Genotypes of offspring _____

Phenotypes of offspring _____

Ratio of phenotypes _____

(3)

- (d) Polydactyly in cats is an inherited condition in which cats have extra toes. The allele for polydactyly is dominant.
- (i) In a population, 19% of cats had extra toes. Use the Hardy-Weinberg equation to calculate the frequency of the recessive allele for this gene in this population.
Show your working.

Answer = _____

(2)

- (ii) Some cat breeders select for polydactyly. Describe how this would affect the frequencies of the homozygous genotypes for this gene in their breeding populations over time.

(1)

(Total 8 marks)

Q10.

Some populations of flies are becoming resistant to insecticides intended to kill them.

Scientists developed a method for finding out whether a fly was carrying a recessive allele, **r**, that gives resistance to an insecticide. The dominant allele, **R**, of this gene does not give resistance.

The scientists:

- crossed flies with genotype **RR** with flies with genotype **rr**
- obtained DNA samples from the parents and offspring
- used the same restriction endonuclease enzymes on each sample, to obtain DNA fragments.

- (a) Explain why the scientists used the same restriction endonuclease enzymes on each DNA sample.

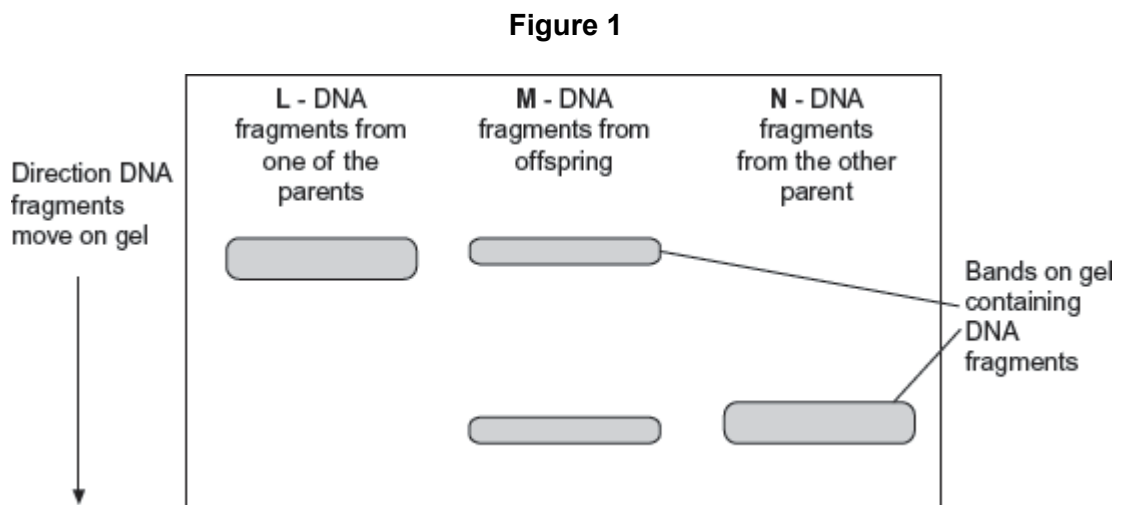
(2)

The scientists added two different primers to each sample of DNA fragments for the polymerase chain reaction (PCR).

- Primer A3 only binds to a 195 base-pair fragment from allele **r**.
- Primer A4 only binds to a 135 base-pair fragment from allele **R**.

The scientists separated the DNA fragments produced by the PCR on a gel where shorter fragments move further in a given time.

Their results are shown in **Figure 1**.



- (b) Explain why primer A3 and primer A4 only bind to specific DNA fragments.

(2)

- (c) Use all the information given to explain the results in **Figure 1**.

(3)

- (d) The scientists wanted to know on which chromosome the gene with alleles **R** and **r** was located. From the flies with genotype **RR**, they obtained cells that were in mitosis and added a labelled DNA probe specific for allele **R**. They then looked at the cells under an optical microscope.

Explain why they used cells that were in mitosis.

(2)

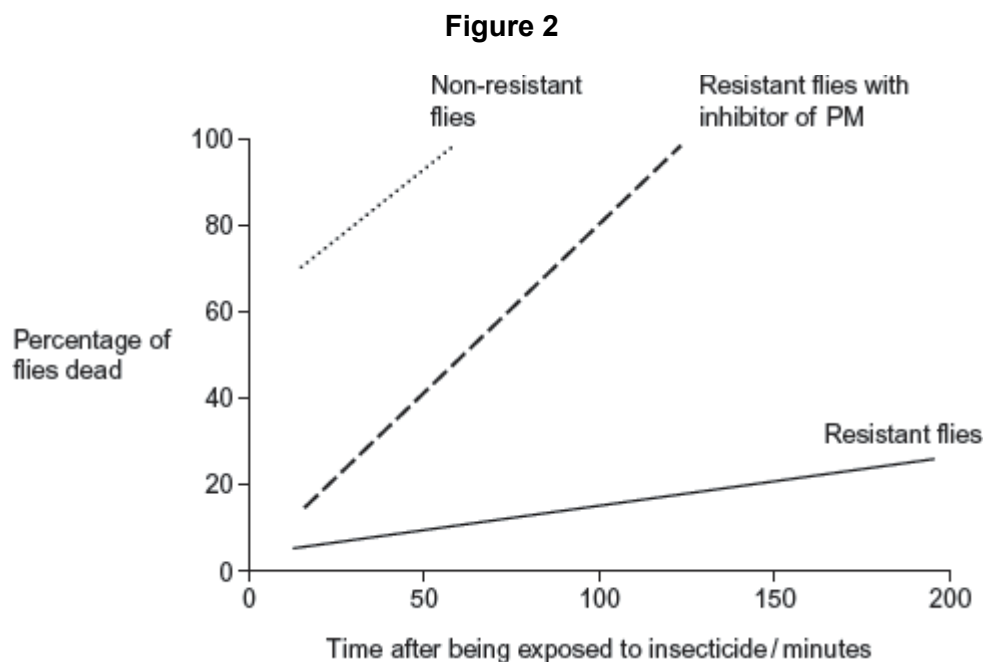
- (e) Another group of scientists thought that pesticide resistance in some flies was related to increased activity of an enzyme called P450 monooxygenase (PM). This enzyme breaks down insecticides.

The scientists obtained large numbers of resistant and non-resistant flies. They then set up the following experiments.

- Non-resistant flies exposed to insecticide.
- Resistant flies exposed to insecticide.
- Resistant flies treated with an inhibitor of PM and then exposed to insecticide.

They then determined the percentage of flies that were dead at different times after being exposed to insecticide.

Figure 2 shows their results.



- (i) Explain why the scientists carried out the control experiment with the non-resistant flies.

(2)

- (ii) The scientists concluded that the resistance of the flies to the insecticide is partly due to increased activity of PM but other factors are also involved.

Explain how these data support this conclusion.

(4)

(Total 15 marks)

Q11.

In birds, **males are XX** and **females are XY**.

- (a) Use this information to explain why recessive, sex-linked characteristics are more common in female birds than in male birds.

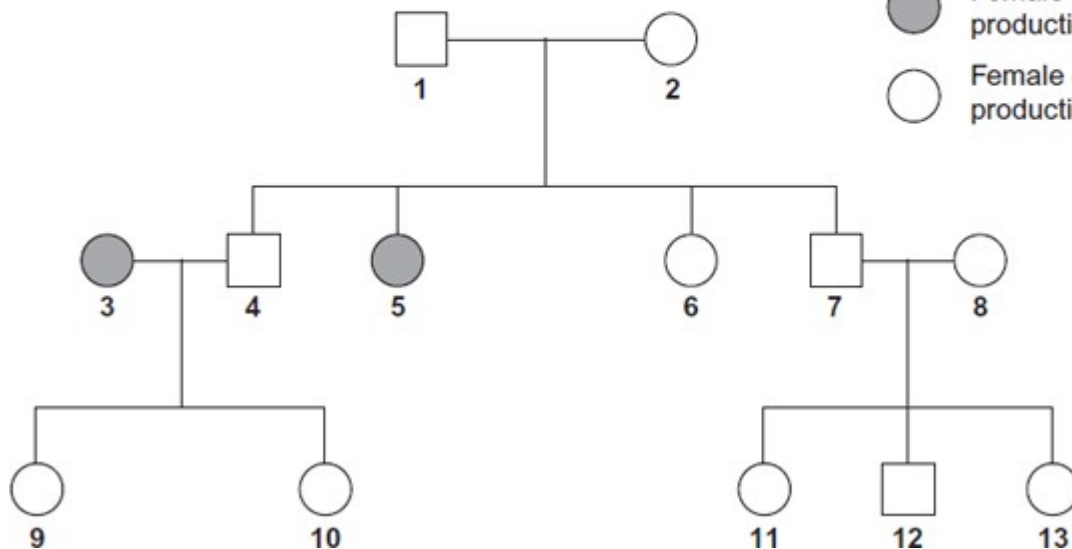
(1)

- (b) In chickens, a gene on the X chromosome controls the rate of feather production. The allele for slow feather production, **F**, is dominant to the allele for rapid feather production, **f**. The following figure shows the results produced from crosses carried out by a farmer.

Male chickens are XX
Female chickens are XY

Key

-  Male - rapid feather production
-  Male - slow feather production
-  Female - rapid feather production
-  Female - slow feather production



- (i) Explain **one** piece of evidence from the figure which shows that the allele for rapid feather production is recessive.

(2)

- (ii) Give all the possible genotypes of the following chickens from the figure.

Chicken 5 _____

Chicken 7 _____

(2)

- (iii) A cross between two chickens produced four offspring. Two of these were males with rapid feather production and two were females with slow feather production. Give the genotypes of the parents.

(1)

- (c) Feather colour in one species of chicken is controlled by a pair of codominant alleles which are **not** sex-linked. The allele C^B codes for black feathers and the allele C^W codes for white feathers. Heterozygous chickens are blue-feathered.

On a farm, 4% of the chickens were black-feathered. Use the Hardy-Weinberg equation to calculate the percentage of this population that you would expect to be blue-feathered. Show your working.

Answer _____ %

(3)

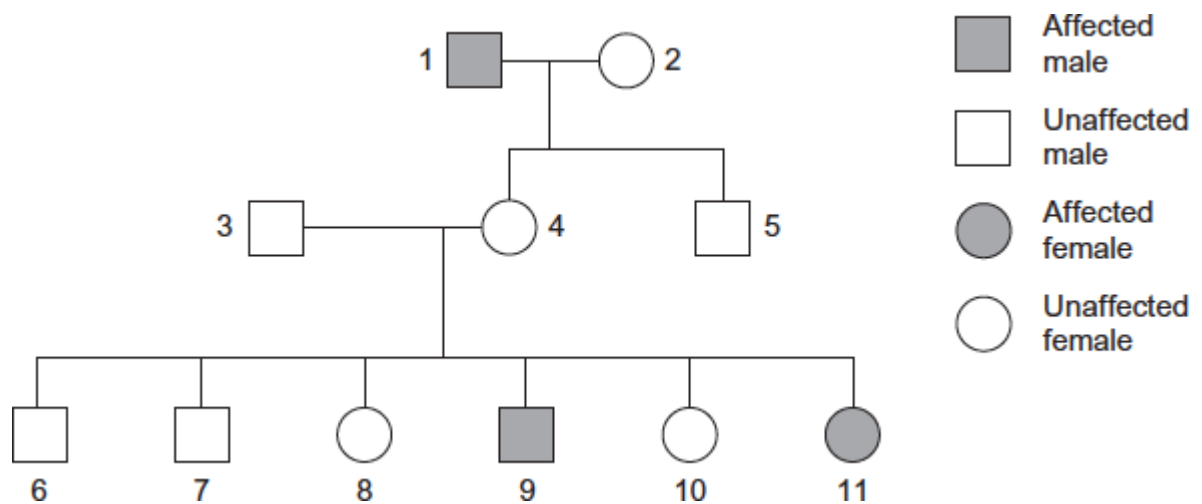
(Total 9 marks)

Q12.

- (a) Explain what is meant by the term phenotype.

(2)

- (b) Tay-Sachs disease is a human inherited disorder. Sufferers of this disease often die during childhood. The allele for Tay-Sachs disease t , is recessive to allele T , present in unaffected individuals. The diagram shows the inheritance of Tay-Sachs in one family.



- (i) Explain **one** piece of evidence from the diagram which proves that the allele for Tay-Sachs disease is recessive.

_____ (2)

(ii) Explain **one** piece of evidence from the diagram which proves that the allele for Tay-Sachs disease is **not** on the X chromosome.

(2)

(c) (i) In a human population, one in every 1000 children born had Tay-Sachs disease. Use the Hardy-Weinberg equation to calculate the percentage of this population you would expect to be heterozygous for this gene. Show your working.

Answer = _____ %

(3)

(ii) The actual percentage of heterozygotes is likely to be lower in future generations than the answer to part (c)(i). Explain why.

(1)

(Total 10 marks)

Q13.

Researchers investigated some characteristics of people from different parts of England. In the north of England they selected 200 people and recorded their phenotypes for three different characteristics.

Their results are shown in the figure below.

Phenotype produced by dominant allele	Number of people	Phenotype produced by recessive allele	Number of people

Tongue roller	131	Non-tongue roller	58
Right-handed	182	Left-handed	14
Straight thumb	142	Hitch-hiker thumb	50

- (a) Calculate the ratio of straight thumb to hitch-hiker thumb in this study.

Ratio = _____

(1)

- (b) The numbers for the tongue rolling and thumb characteristics do not add up to 200. For each characteristic suggest **one** reason why the numbers do **not** add up to 200.

Tongue rolling _____

Thumb _____

(2)

- (c) One student looked at the researchers' results and concluded that 91% of people in the UK are right-handed. Do you agree with this conclusion? Give reasons for your answer.

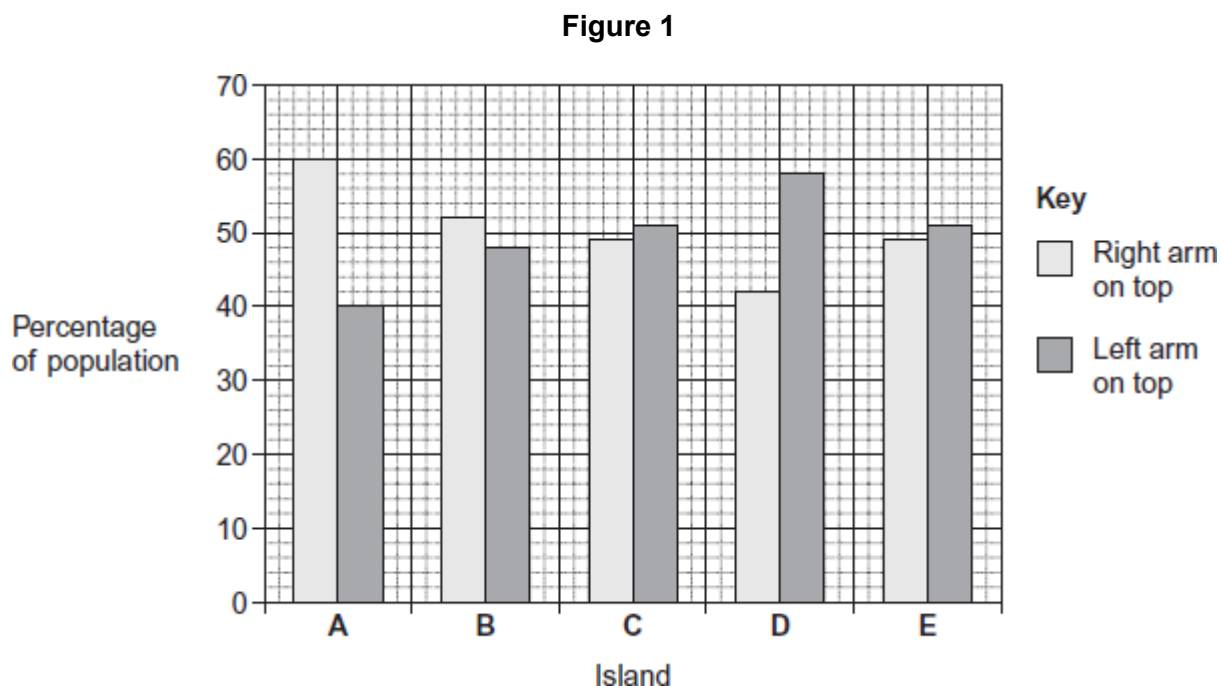
(2)

(Total 5 marks)

Q14.

When most people fold their arms, they either always have their left arm on top, **L**, or always have their right arm on top, **R**. A geneticist investigated this characteristic on five small islands, **A**, **B**, **C**, **D** and **E**.

Her results are shown in **Figure 1**.



On one of the islands she recorded the arm-folding characteristics of parents and their children.

These results are shown in **Figure 2**.

Figure 2

Arm-folding of parents	Arm-folding of the children / %	
	Right arm on top, R	Left arm on top, L
R and R	41	59
R and L	45	55
L and L	44	56

The geneticist concluded that arm-folding is not determined by a single gene with a dominant allele and a recessive allele.

(a) The geneticist investigated arm-folding on five small islands.

(i) Use information from **Figure 1** to describe the results she obtained.

(2)

- (ii) Suggest advantages of using island populations in this investigation.

(2)

- (b) The geneticist concluded that arm-folding is **not** determined by a single gene with a dominant allele and a recessive allele.

Use information from **Figure 2** to explain why she reached this conclusion.

(3)

- (c) In another study, the geneticist investigated arm-folding in genetically identical twins. Data from this study supported her conclusion from the island study.

Suggest the evidence she found that supported her conclusion.

(1)

(Total 8 marks)

Q15.

The fruit fly is a useful organism for studying genetic crosses. Female fruit flies are approximately 2.5 mm long. Males are smaller and possess a distinct black patch on their bodies. Females lay up to 400 eggs which develop into adults in 7 to 14 days. Fruit flies will survive and breed in small flasks containing a simple nutrient medium consisting mainly of sugars.

- (a) Use this information to explain **two** reasons why the fruit fly is a useful organism for studying genetic crosses.

1. _____

2. _____

(2)

- (b) Male fruit flies have the sex chromosomes XY and the females have XX. In the fruit fly, a gene for eye colour is carried on the X chromosome. The allele for red eyes, **R**, is dominant to the allele for white eyes, **r**. The genetic diagram shows a cross between two fruit flies.

- (i) Complete the genetic diagram for this cross.

Phenotypes of parents	red-eyed female		white-eyed male
Genotype of parents	_____	×	_____
Gametes	_____ and _____		_____ and _____
Phenotypes of offspring	red-eyed females	and	red-eyed males
Genotype of offspring	_____		_____

(3)

- (ii) The number of red-eyed females and red-eyed males in the offspring was counted. The observed ratio of red-eyed females to red-eyed males was similar to, but not the same as, the expected ratio. Suggest **one** reason why observed ratios are often **not** the same as expected ratios.

(1)

- (c) Male fruit flies are more likely than female fruit flies to show a phenotype produced by a recessive allele carried on the X chromosome. Explain why.

(2)

(Total 8 marks)

Q16.

In a species of snail, shell colour is controlled by a gene with three alleles. The shell may be brown, pink or yellow. The allele for brown, C^B , is dominant to the other two alleles. The allele for pink, C^P , is dominant to the allele for yellow, C^Y .

- (a) Explain what is meant by a *dominant* allele.

(1)

- (b) Give **all** the genotypes which would result in a brown-shelled snail.

(1)

- (c) A cross between two pink-shelled snails produced only pink-shelled and yellow-shelled snails. Use a genetic diagram to explain why.

(3)

- (d) The shells of this snail may be unbanded or banded. The absence or presence of bands is controlled by a single gene with two alleles. The allele for unbanded, B , is dominant to the allele for banded, b .

A population of snails contained 51% unbanded snails. Use the Hardy-Weinberg equation to calculate the percentage of this population that you would expect to be heterozygous for this gene. Show your working.

Answer _____ %

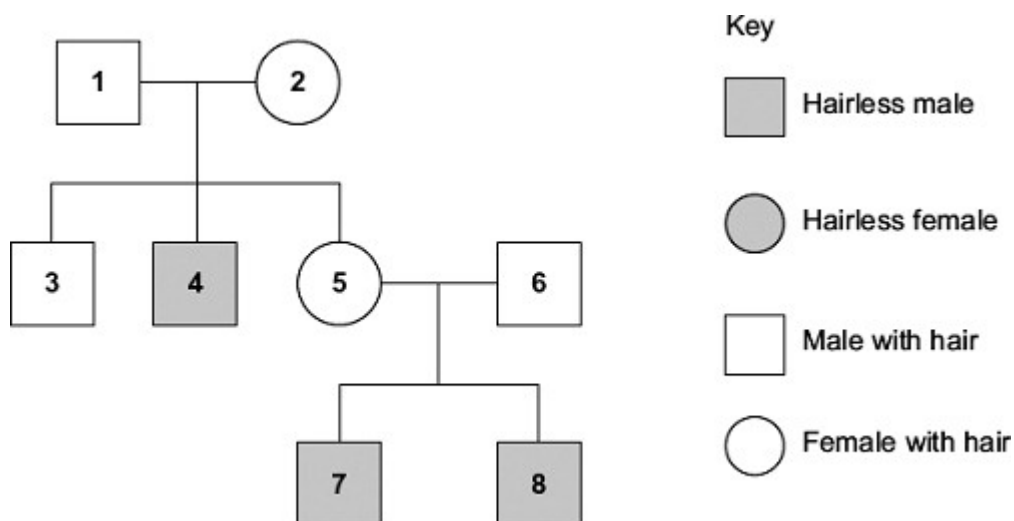
(3)

(Total 8 marks)

Q17.

A single gene controls the presence of hair on the skin of cattle. The gene is carried on the X chromosome. Its dominant allele causes hair to be present on the skin and its recessive allele causes hairlessness.

The diagram shows the pattern of inheritance of these alleles in a group of cattle.



(a) Use evidence from the diagram to explain

(i) that hairlessness is caused by a recessive allele

(2)

(ii) that hairlessness is caused by a gene on the X chromosome.

(1)

(b) What is the probability of the next calf born to animals **5** and **6** being hairless? Complete the genetic diagram to show how you arrived at your answer.

Phenotypes of parents

Female with hair

Male with hair

Genotypes of parents

Gametes

Genotypes of offspring _____

Phenotypes of offspring _____

Probability of next calf being hairless _____

(4)
(Total 7 marks)

Q18.

A breeder crossed a black male cat with a black female cat on a number of occasions. The female cat produced 8 black kittens and 4 white kittens.

- (a) (i) Explain the evidence that the allele for white fur is recessive.

(1)

- (ii) Predict the likely ratio of colours of kittens born to a cross between **this** black male and a white female.

(1)

- (b) The gene controlling coat colour has three alleles. The allele **B** gives black fur, the allele **b** gives chocolate fur and the allele **bⁱ** gives cinnamon fur.

- Allele **B** is dominant to both allele **b** and **bⁱ**.
- Allele **b** is dominant to allele **bⁱ**.

- (i) Complete the table to show the phenotypes of cats with each of the genotypes shown.

Genotype	Phenotype
Bbⁱ	
bbⁱ	
Bb	

(1)

- (ii) A chocolate male was crossed several times with a black female.

They produced

- 11 black kittens

- 2 chocolate kittens
- 5 cinnamon kittens.

Using the symbols in part (b), complete the genetic diagram to show the results of this cross.

<i>Parental phenotypes</i>	Chocolate male	Black female
<i>Parental genotypes</i>	_____	_____
<i>Gametes</i>	_____	_____

<i>Offspring genotypes</i>	_____	_____	_____
<i>Offspring phenotypes</i>	Black	Chocolate	Cinnamon

(3)

- (iii) The breeder had expected equal numbers of chocolate and cinnamon kittens from the cross between the chocolate male and black female. Explain why the actual numbers were different from those expected.

(1)

- (iv) The breeder wanted to produce a population of cats that would all have chocolate fur. Is this possible? Explain your answer.

(2)

(Total 9 marks)

Q19.

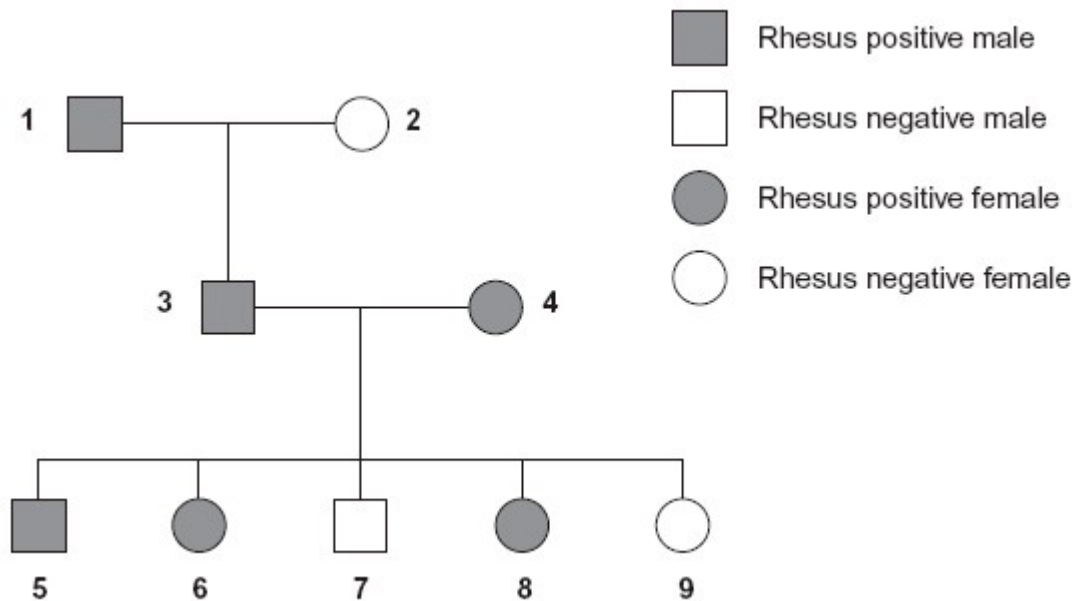
- (a) (i) Explain what is meant by a **recessive** allele.

(1)

- (ii) Explain what is meant by **codominant** alleles.

(1)

- (b) The Rhesus blood group is genetically controlled. The gene for the Rhesus blood group has two alleles. The allele for Rhesus positive, **R**, is dominant to that for Rhesus negative, **r**. The diagram shows the inheritance of the Rhesus blood group in one family.



- (i) Explain **one** piece of evidence from the diagram which shows that the allele for Rhesus positive is dominant.

(2)

- (ii) Explain **one** piece of evidence from the diagram which shows that the gene is **not** on the X chromosome.

(2)

- (c) Sixteen percent of the population of Europe is Rhesus negative. Use the Hardy-Weinberg equation to calculate the percentage of this population that you would expect to be heterozygous for the Rhesus gene.

Show your working.

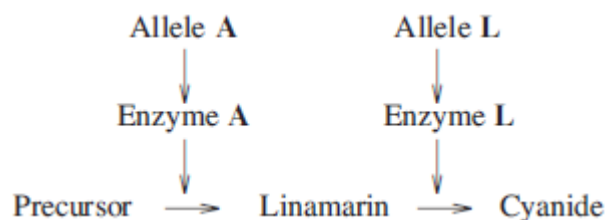
Answer _____

(3)

(Total 9 marks)

Q20.

Cyanide is a poisonous substance. Cyanogenic clover plants produce cyanide when their tissues are damaged. The ability to produce cyanide is controlled by genes at loci on two different chromosomes. The dominant allele, **A**, of one gene controls the production of an enzyme which converts a precursor to linamarin. The dominant allele, **L**, of the second gene controls the production of an enzyme which converts linamarin to cyanide. This is summarised in the diagram.



- (a) Acyanogenic clover plants cannot produce cyanide. Explain why a plant with the genotype **aaLI** cannot produce cyanide.

(1)

(b) A clover plant has the genotype **AaLl**.

(i) Give the genotypes of the male gametes which this plant can produce.

_____ (1)

(ii) Explain how meiosis results in this plant producing gametes with these genotypes.

_____ (2)

(c) Two plants, heterozygous for both of these pairs of alleles, were crossed. What proportion of the plants produced from this cross would you expect to be acyanogenic but able to produce linamarin? Use a genetic diagram to explain your answer.

(3)

In an investigation, cyanogenic and acyanogenic plants were grown together in pots. Slugs were placed in each pot and records were kept of the number of leaves damaged by the feeding of the slugs over a period of 7 days. The results are shown in **Table 1**.

Table 1

	Undamaged	Damaged
Cyanogenic plants	160	120
Acyanogenic plants	88	192

(d) A χ^2 test was carried out on the results.

(i) Suggest the null hypothesis that was tested.

(1)

- (ii) χ^2 was calculated. When this value was looked up in a table, it was found to correspond to a probability of less than 0.05. What conclusion can you draw from this?

(3)

A second investigation was carried out in a field of grass which had been undisturbed for many years. **Table 2** shows the population density of slugs and the numbers of cyanogenic and acyanogenic clover plants at various places in the field.

Table 2

Population density of slugs	Number of acyanogenic clover plants per m ²	Number of cyanogenic clover plants per m ²
Very low	26	10
Low	17	26
High	0	10
Very high	0	5

- (e) Explain the proportions of the two types of clover plant in different parts of the field.

(4)

Q21.

- (a) Meiosis results in cells that have the haploid number of chromosomes and show genetic variation. Explain how.

(6)

- (b) In mice, two genes affecting coat colour are on different chromosomes. One gene controls whether there is any black pigment in the hairs. The dominant allele of this gene, **B**, results in black fur. The recessive allele, **b**, results in white fur. The second gene controls banding of the fur. The dominant allele, **A**, causes a yellow band to develop on each hair. The resulting coat colour is called agouti. The recessive allele, **a**, results in hairs with no bands on them. This gene has no effect on mice with white fur; white mice do not develop bands, even if they have the **A** allele.

Breeders performed many crosses in which agouti mice were crossed with white mice, homozygous for both genes. They expected agouti, black and white mice in the offspring in a 1 : 1 : 2 ratio.

- (i) Complete the genetic diagram to show how this ratio of phenotypes would be produced.

Parental phenotypes	Agouti	White
Parental genotypes		
Gamete genotypes		
Offspring genotypes		
Offspring phenotypes		

(4)

- (ii) The actual numbers of offspring with each phenotype were

Agouti	34
Black	35
White	51

The χ^2 test can be used to test the hypothesis that there is no significant difference between these results and the expected 1 : 1 : 2 ratio. Complete the table to calculate the value of χ^2 for these results.

Colour of offspring	Observed (O)	Expected (E)	(O - E)	(O - E) ²	$\frac{(O - E)^2}{E}$
Agouti	34				
Black	35				
White	51				
			$\sum \frac{(O - E)^2}{E} =$		

(2)

- (iii) The table shows values for χ^2 at different levels of probability and for different degrees of freedom.

Degrees of freedom	Probability, p				
	0.2	0.1	0.05	0.02	0.01
1	1.64	2.71	3.84	5.41	6.64
2	3.22	4.61	5.99	7.82	9.21
3	4.64	6.25	7.82	9.84	11.35
4	5.99	7.78	9.49	11.67	13.28
5	7.29	9.24	11.07	13.39	15.09

What should the breeders conclude about the significance of their results? Explain your answer.

Q22.

- (a) The guinea pig, *Cavia porcellus*, is a small mammal. Complete the table to show the classification of the guinea pig.

Kingdom	
	Chordata
	Mammalia
	Rodentia
Family	Caviidae
Genus	
Species	

(2)

- (b) In South America, there are several species of guinea pig. They are thought to have arisen by sympatric speciation.
Explain how sympatric speciation may have occurred.

(3)

- (c) In guinea pigs, hair length and hair colour are controlled by two genes on different chromosomes. The hair may be either long or short and its colour either black or brown.

A male guinea pig and a female guinea pig both had short, black hair. The male was homozygous for hair length, and the female was homozygous for hair colour. Repeated crossings of these two guinea pigs resulted in offspring of four different genotypes, all of which had short, black hair.

Complete the genetic diagram to explain these results. Write in the box the symbols you will use to represent the alleles.

Allele for short hair = _____	Allele for long hair = _____
-------------------------------	------------------------------

Allele for black hair = _____	Allele for brown hair = _____
-------------------------------	-------------------------------

Parental phenotypes	Male Short, black hair	Female Short, black hair
Parental genotypes	_____	_____
Gamete genotypes	_____	_____

Offspring genotypes	_____
Offspring phenotypes	Short, black hair

(4)

- (d) In another investigation, the same female guinea pig was twice mated with another male which had long, brown hair. Of the 14 offspring, 10 had short, black hair and 4 had long, black hair. The investigators expected equal numbers of offspring with these two phenotypes. They used a χ^2 test to determine whether the observed results fitted the expected 1:1 ratio.

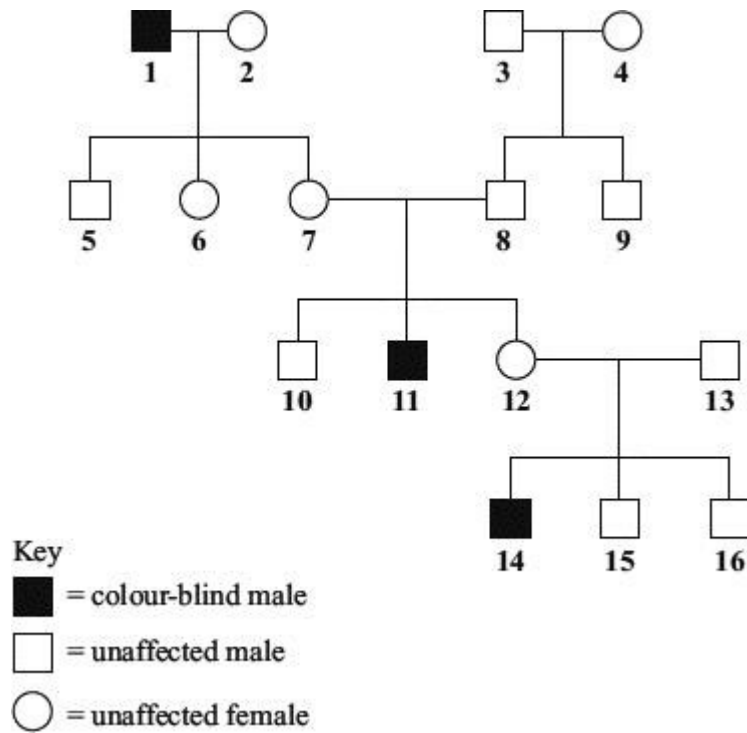
Give a suitable null hypothesis for the investigation.

(1)

(Total 10 marks)

Q23.

Red-green colour blindness is caused by a mutation in the gene coding for one of the opsin proteins which are needed for colour vision. The diagram shows the inheritance of red-green colour blindness in one family.



Person **12** is pregnant with her fourth child. What is the probability that this child will be a male with red-green colour blindness? Explain your answer by drawing a genetic diagram. Use the following symbols

X^R = an X chromosome carrying an allele for normal colour vision

X^r = an X chromosome carrying an allele for red-green colour blindness

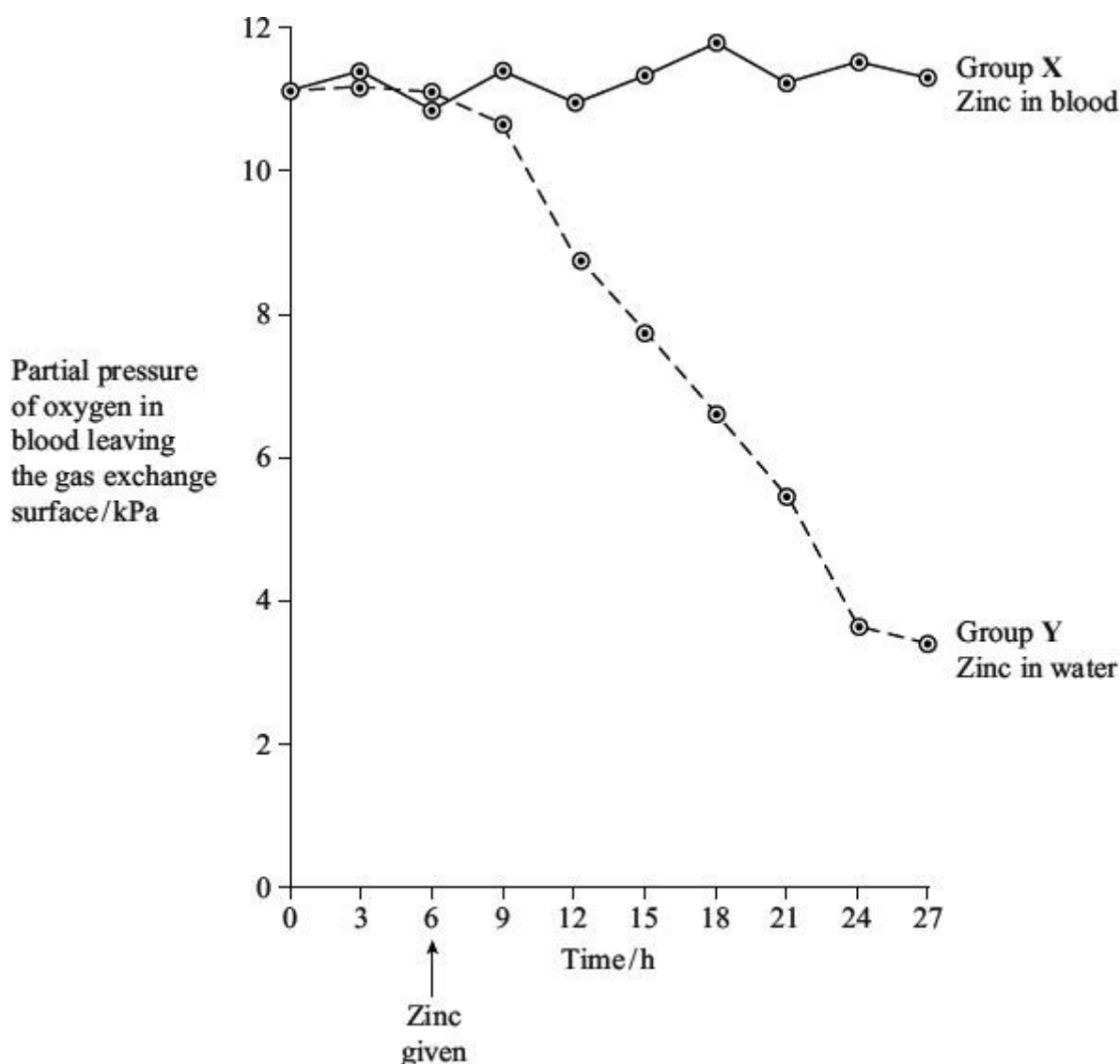
Y = a Y chromosome

Q24.

Ions of metals such as zinc often pollute rivers. The effect of zinc ions on gas exchange and respiration in fish was investigated. Fish were kept in tanks of water in a laboratory.

The fish in one group (X) had a solution of a zinc compound injected directly into their blood and were then put in a tank of zinc-free water. A second group (Y) was not injected but had the solution of the zinc compound added to the water in the tank.

The partial pressure of oxygen in the blood of both groups of fish was then monitored. The results are shown in the graph.



- (a) During this investigation, the water temperature in the tanks was kept constant. Explain why changes in the water temperature might lead to the results of the investigation being unreliable.

-
- (b) The results from the two groups were compared using a statistical test.
- (i) Suggest a null hypothesis that could be tested.
-
- (ii) Explain why it is important to use a statistical test in analysing the results of this investigation.
-
-
-
-
- (c) Two suggestions were made to explain the results shown in the graph.
- A** Zinc ions reduce the rate at which oxygen is taken up from the water and passes into the blood.
- B** Zinc ions reduce the ability of haemoglobin to transport oxygen.
- Which of these suggestions is the more likely? Explain the evidence from the graph that supports your answer.
-
-
-
-
- (d) During the investigation, the pH of the blood was also monitored. It decreased in group Y. Suggest an explanation for this decrease in pH.
-
-
-
-
-
-
- (e) Leaves were collected from sycamore trees growing in a polluted wood and the

concentration of some metal ions in samples of these leaves was measured. Woodlice were then fed with the leaves. After 20 weeks, the concentration of the ions in the bodies of the woodlice was measured. Some of the results are shown in the table.

	Concentration of ions / $\mu\text{g g}^{-1}$			
	Copper	Cadmium	Zinc	Lead
Leaves	52	26	1430	908
Woodlice	1130	525	1370	132

- (i) Which of the elements shown in the table is concentrated most by the woodlice? Use suitable calculations to support your answer.

(2)

- (ii) Suggest what happens to most of the lead ions in the leaves eaten by the woodlice.

(1)

- (iii) Explain the difference in the copper ion concentration between the leaves and the woodlice.

(2)

- (f) Yorkshire fog is a species of grass. Two varieties of Yorkshire fog were studied. One variety was tolerant to arsenic, while the other variety was not. In a series of investigations, it was found that

- Arsenic-tolerant plants grow in soil which contains a high concentration of arsenic.
- Arsenic-tolerant plants growing in soil containing high concentrations of arsenic and phosphorus-containing compounds have very low concentrations of arsenic in their cells. They also have low concentrations of phosphates in their cells. Arsenic and phosphorus are chemically similar.
- Plants that are not tolerant to arsenic grow poorly on soil which has a high

concentration of both arsenic and phosphorus-containing compounds.

- Tolerance to arsenic in Yorkshire fog is caused by a single gene with the allele, **a**, for tolerance recessive to the allele, **A**, for non-tolerance.

(i) What caused the allele for tolerance to first arise?

(1)

(ii) Give **two** functions of phosphates in plant cells.

1.

2.

(2)

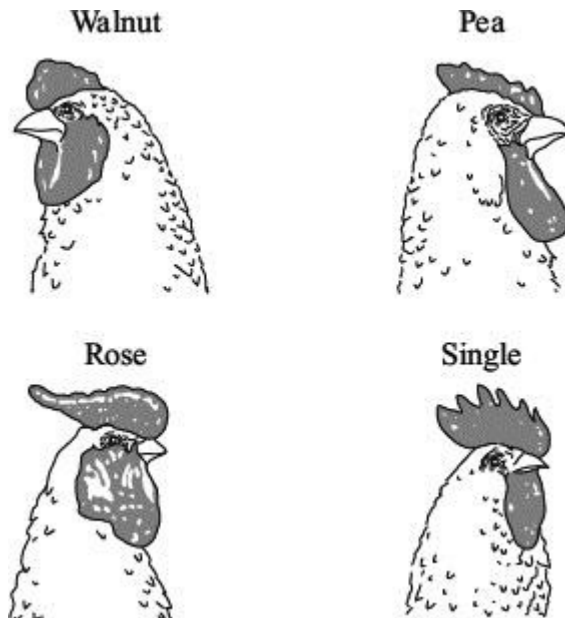
(iii) Arsenic-tolerant Yorkshire fog plants are very rare in areas with low concentrations of arsenic in the soil, even where the soil has a high concentration of phosphate. Explain why they are unable to compete in these conditions with plants that are not tolerant to arsenic.

(3)

(Total 20 marks)

Q25.

Chickens have a structure on their heads called a comb. The diagram shows four types of comb: walnut, pea, rose and single.



Two genes control the type of comb; each gene has a dominant and a recessive allele. The two genes are inherited independently, but interact to produce the four types of comb.

Genotype	Phenotype
A- B-	Walnut
A- bb	Pea
aa B-	Rose
aa bb	Single

The symbol - indicates that either the dominant allele or recessive allele could be present

- (a) A male with a pea comb, heterozygous for gene A, was crossed with a rose-combed female, heterozygous for gene B. Complete the genetic diagram to show the offspring expected from this cross.

Phenotypes of parents	Pea comb	Rose comb
Genotypes of parents	_____	_____
Gametes formed	_____	_____

Offspring genotypes	_____
Ratio of offspring phenotypes	_____

(3)

- (b) Chickens with rose or single combs made up 36% of one population. Assuming the

conditions of the Hardy-Weinberg equilibrium apply, calculate the frequency of allele **a** in this population. Show how you arrived at your answer.

Frequency of allele **a** = _____

(2)

(Total 5 marks)

Q26.

Human ABO blood groups are determined by the presence or absence of two antigens (A and B) on the plasma membrane of the red blood cells. The inheritance of these blood groups is controlled by three alleles:

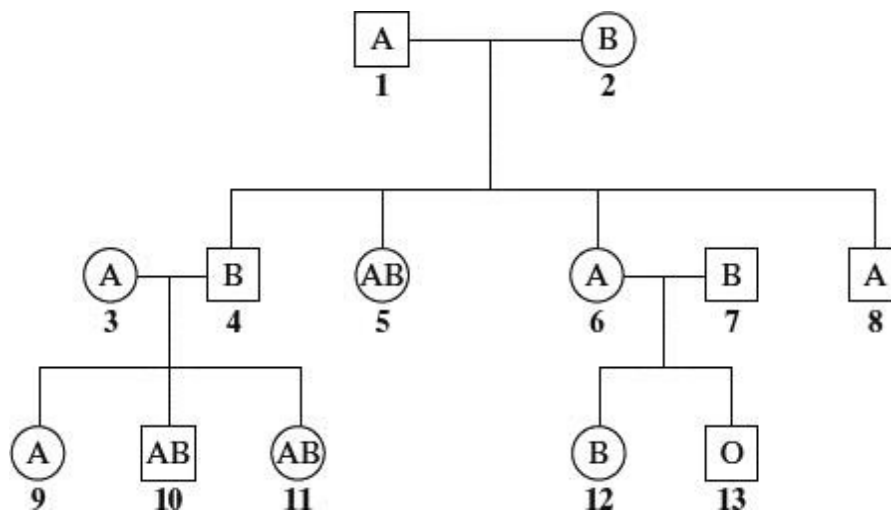
I^A – determines the production of antigen A

I^B – determines the production of antigen B

I^O – determines the production of no antigen

Alleles I^A and I^B are codominant. Allele I^O is recessive to both.

The pedigree shows the pattern of inheritance of these blood groups in a family over three generations.



- (a) (i) How many antigen-determining alleles will be present in a white blood cell?
Give a reason for your answer.

(1)

- (ii) Which antigen or antigens will be present on the plasma membranes of red blood cells of individual **5**?

(1)

- (b) If individuals **6** and **7** were to have another child, what is the probability that this child would be male and blood group A? Explain your answer.

(3)

(Total 5 marks)

Q27.

- (a) The control of water balance in the body involves negative feedback.

- (i) Describe what is meant by *negative feedback*.

(1)

- (ii) Water is removed from the body via the kidneys. Give **two** other ways in which water is removed from the body.

1. _____

2. _____

(2)

- (iii) Name the part of the brain which acts as the coordinator in the control of water balance.

(1)

- (b) **Figure 1** shows the cells lining the collecting duct in a human kidney. ADH molecules bind to the receptor proteins and this triggers the vesicles containing aquaporins to bind with the plasma membrane next to the lumen. **Figure 2** shows an aquaporin which is a large channel protein.

Figure 1

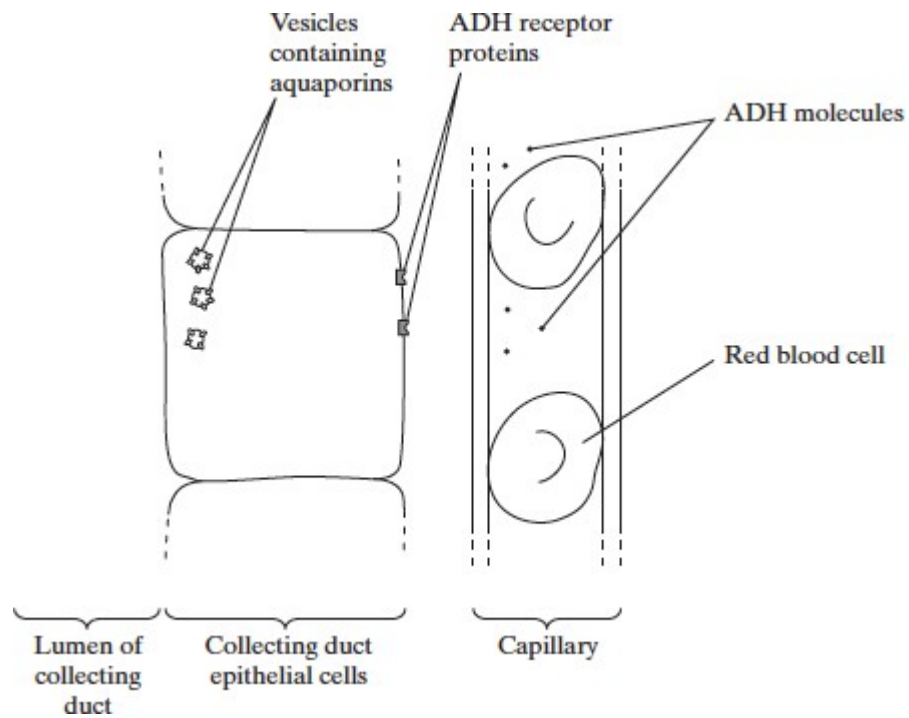
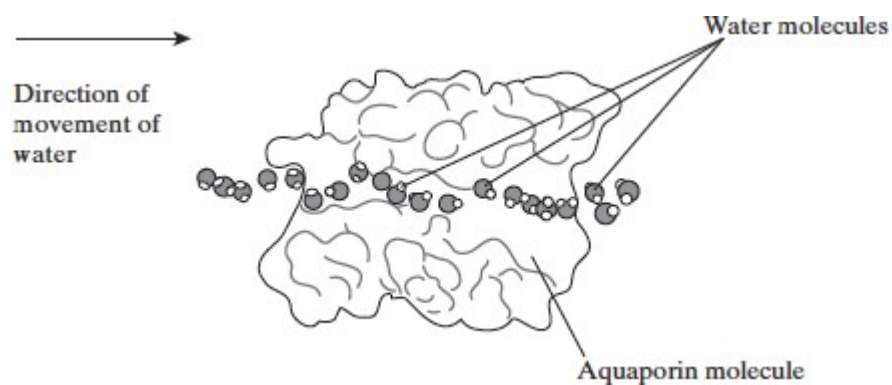


Figure 2



- (i) From which gland is ADH released?

(1)

- (ii) Use the information given to explain how ADH increases the movement of water from the lumen of the collecting duct into the blood.

(4)

- (c) The gene for the ADH receptor proteins is found on the X chromosome. One allele of this gene causes a non-functioning receptor protein to be made. This allele is recessive and is one cause of the condition called diabetes insipidus.

- (i) What would be the most obvious symptom of diabetes insipidus?

(1)

- (ii) Suggest why diabetes insipidus is more common in males.

(2)

- (iii) A recessive allele which has harmful effects is able to reach a higher frequency in a population than a harmful dominant allele. Explain how.

(3)

(Total 15 marks)

Q28.

In fruit flies, the allele for grey body, **G**, is dominant to the allele for ebony body, **g**, and the allele for normal wings, **N**, is dominant to the allele for vestigial wings, **n**. Vestigial-winged flies, heterozygous for grey body colour, were crossed with ebony-bodied flies, heterozygous for normal wings.

Complete the genetic diagram to show the genotypes and phenotypes in this cross.

Parental phenotypes Grey body, vestigial wings Ebony body, normal wings

Parental genotypes

Gamete genotypes

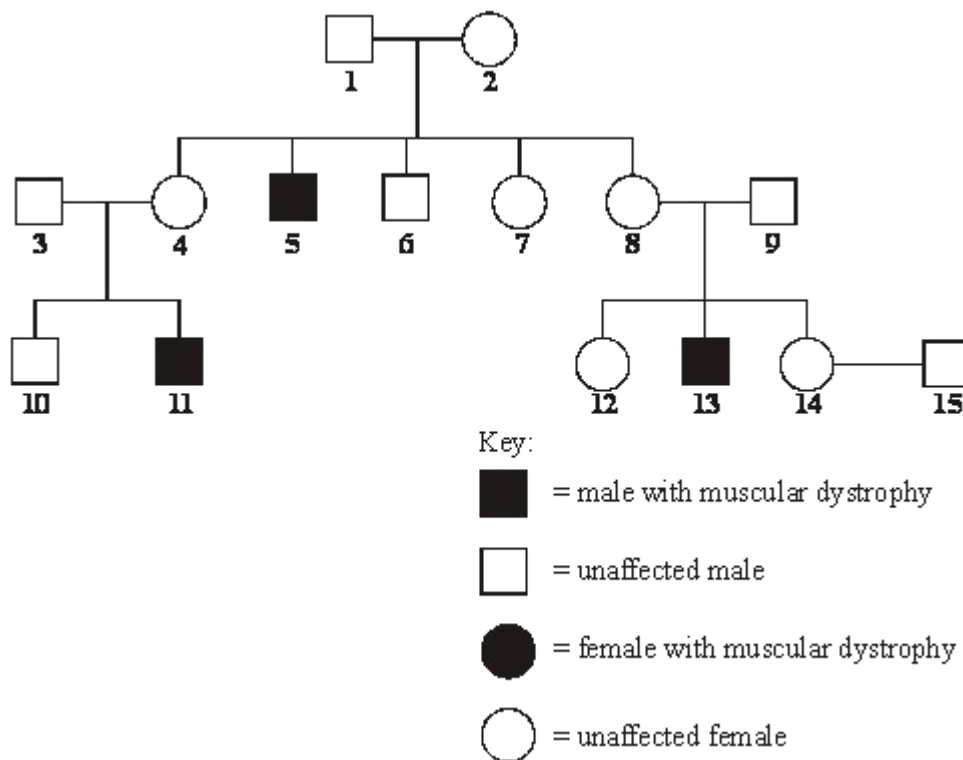
Offspring genotypes

Offspring phenotypes

(Total 4 marks)

Q29.

Duchenne muscular dystrophy is a sex-linked inherited condition which causes degeneration of muscle tissue. It is caused by a recessive allele. The diagram shows the inheritance of muscular dystrophy in one family.



(a) Give evidence from the diagram which suggests that muscular dystrophy is

(i) sex-linked; _____

(1)

(ii) caused by a recessive allele. _____

(1)

(b) Using the following symbols,

X^D = an X chromosome carrying the normal allele

X^d = an X chromosome carrying the allele for muscular dystrophy

Y = a Y chromosome

give **all** the possible genotypes of each of the following persons.

5 _____

6 _____

7 _____

8 _____

(2)

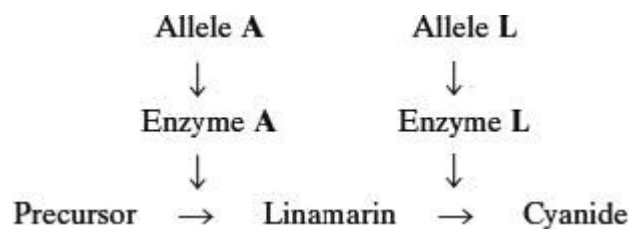
(c) A blood test shows that person **14** is a carrier of muscular dystrophy. Person **15** has recently married person **14** but as yet they have had no children. What is the probability that their first child will be a male who develops muscular dystrophy?

(1)

(Total 5 marks)

Q30.

Cyanide is a poisonous substance. Cyanogenic clover plants produce cyanide when their tissues are damaged. The ability to produce cyanide is controlled by genes at loci on two different chromosomes. The dominant allele, **A**, of one gene controls the production of an enzyme which converts a precursor to linamarin. The dominant allele, **L**, of the second gene controls the production of an enzyme which converts linamarin to cyanide. This is summarised in the diagram.



(a) Acyanogenic clover plants cannot produce cyanide. Explain why a plant with the genotype **aaLl** cannot produce cyanide.

(1)

(b) A clover plant has the genotype **AaLl**.

(i) Give the genotypes of the male gametes which this plant can produce.

_____ (1)

(ii) Explain how meiosis results in this plant producing gametes with these genotypes.

_____ (2)

(c) Two plants, heterozygous for both of these pairs of alleles, were crossed. What proportion of the plants produced from this cross would you expect to be acyanogenic but able to produce linamarin? Use a genetic diagram to explain your answer.

(3)

In an investigation, cyanogenic and acyanogenic plants were grown together in pots. Slugs were placed in each pot and records were kept of the number of leaves damaged by the feeding of the slugs over a period of 7 days. The results are shown in **Table 1**.

Table 1

	Undamaged	Damaged
Cyanogenic plants	160	120
Acyanogenic plants	88	192

(d) A χ^2 test was carried out on the results.

(i) Suggest the null hypothesis that was tested.

(1)

- (ii) χ^2 was calculated. When this value was looked up in a table, it was found to correspond to a probability of less than 0.05. What conclusion can you draw from this?

(3)

A second investigation was carried out in a field of grass which had been undisturbed for many years. **Table 2** shows the population density of slugs and the numbers of cyanogenic and acyanogenic clover plants at various places in the field.

Table 2

Population density of slugs	Number of acyanogenic clover plants per m ²	Number of cyanogenic clover plants per m ²
Very low	26	10
Low	17	26
High	0	10
Very high	0	5

- (e) Explain the proportions of the two types of clover plant in different parts of the field.

(4)

Q31.

The inheritance of body colour in fruit flies was investigated. Two fruit flies with grey bodies were crossed. Of the offspring, 152 had grey bodies and 48 had black bodies.

- (a) Using suitable symbols, give the genotypes of the parents. Explain your answer.

Genotypes _____

Explanation _____

(2)

- (b) Explain why a statistical test should be applied to the data obtained in this investigation.

(2)

- (c) A species of insect, only found on a remote island, has a characteristic controlled by a pair of codominant alleles, C^M and C^N .

- (i) What is meant by *codominant*?

(1)

- (ii) There were 500 insects in the total population. In this population, 300 insects had the genotype $C^M C^M$, 150 had the genotype $C^M C^N$ and 50 had the genotype $C^N C^N$. Calculate the actual frequency of the allele C^N by using these figures. Show your working.

Answer _____

(2)

- (iii) Use your answer to (ii) and the Hardy-Weinberg equation to calculate the number of insects that would be **expected** to have the genotype $C^N C^N$.

Answer _____

(3)

(Total 10 marks)

Q32.

Most tigers have fur that is orange with black stripes. The orange colour is controlled by a single gene. The dominant allele, **T**, leads to the production of orange fur and the recessive allele **t** leads to the production of white fur. The black stripes are controlled by a different gene. The dominant allele of this gene, **A**, leads to the production of stripes and the recessive allele, **a**, leads to the production of unstriped fur. A tiger with white fur and no stripes is called a snowy tiger.

- (a) What is meant by a *recessive* allele?

(1)

- (b) A tiger with orange, striped fur was heterozygous for the gene for coat colour and for the gene for stripes. It was mated with a snowy tiger. Complete the genetic diagram.

Orange, striped tiger × Snowy tiger

Parental genotypes

Genotypes of gametes

Genotypes of offspring

Phenotypes of offspring

(4)

- (c) Snowy tigers inhabit the same grasslands as orange, striped tigers. These grasslands are dominated by very tall grass plants. Snowy tigers are less successful hunters. Suggest why.

(2)

(Total 7 marks)

Q33.

One form of baldness in humans is controlled by two alleles, **B** and **b**, of a single gene. This gene is not on the X chromosome but the expression of the gene is affected by the sex of a person.

Men who are **BB** or **Bb** will become bald. Men who are **bb** will not become bald.

Women who are **BB** will become bald. Women who are **Bb** or **bb** will not become bald.

One type of colour blindness is controlled by a sex-linked gene, found on the X chromosome. The dominant allele **X^A** leads to normal colour vision and the recessive allele **X^a** leads to colour blindness.

- (a) (i) Give all the possible genotypes of a bald man who has normal colour vision.

(1)

- (ii) Give all the possible genotypes of a woman who will not become bald and who carries one allele for colour blindness.

(1)

- (b) A mother and a father are both heterozygous for the gene for baldness. The father has normal colour vision and the mother is heterozygous for the gene for colour blindness. Complete the genetic diagram to show the probability of a son of this couple being colour blind but not becoming bald.

Father

Mother

Genotypes of parents

Gametes

Genotypes of sons

Probability of son being colour blind but not becoming bald _____

(4)

(Total 6 marks)

Q34.

Chickens have a structure called a comb on their heads. The drawings show two types of comb.



Pea comb



Single comb

The shape of the comb is controlled by two alleles of one gene. The allele for pea comb, **A**, is dominant to the allele for single comb, **a**.

The colour of chicken eggs is controlled by two alleles of a different gene. The allele for blue eggs, **B**, is dominant to the allele for white eggs, **b**.

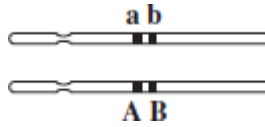
The genes for comb shape and egg colour are situated on the same chromosome.

A farmer crossed a male chicken with the genotype **AaBb** with a female chicken that had a single comb and produced white eggs.

- (a) What was the genotype of the female parent?

(1)

The diagram shows how the alleles of the genes were arranged on the chromosomes of the male parent.



- (b) Which **two** genotypes will be most frequent in the offspring?

_____ (1)

- (c) The farmer could identify which of the female offspring from this cross would eventually produce blue eggs. Explain how.

_____ (2)

- (d) Genes **A** and **B** are close together on the chromosome. This is important when trying to identify which of the female offspring would produce blue eggs. Explain why.

_____ (2)

- (e) Suggest **two** environmental factors which are likely to affect egg production.

1. _____
2. _____ (2)

In chickens it is the males which are XX and the females which are XY.

- (f) A gene on the X chromosome controls the rate of feather production. The allele for slow feather production, **F**, is dominant to the allele for rapid feather production, **f**.

A farmer made a cross between two chickens with known genotypes. He chose these chickens so that he could tell the sex of the offspring soon after they hatched by looking at their feathers.

Which of the crosses shown in the table did he make? Explain your answer.

Cross	Genotype of male parent	Genotype of female parent
A	$X^F X^F$	$X^f Y$

B	$X^F X^f$	$X^f Y$
C	$X^f X^f$	$X^F Y$
D	$X^F X^f$	$X^F Y$

Answer _____

Explanation _____

(3)

- (g) Female chickens are more likely than male chickens to show recessive sex-linked characteristics. Explain why.

(3)

(Total 14 marks)

Q35.

Hair type in dachshund dogs is controlled by two genes which are on different chromosomes.

Dogs with the **H** allele have wiry hair and dogs with the genotype **hh** have non-wiry hair.

The length of wiry hair is always the same. Dogs with non-wiry hair have either long or short hair. The length of non-wiry hair is controlled by another gene. Dogs with the **D** allele have short hair and those with the genotype **dd** have long hair.

- (a) Give all the possible genotypes for dachshunds with non-wiry, short hair.

(1)

- (b) What type of interaction is occurring between the two genes? Explain your answer.

(2)

- (c) A wiry-haired male with the genotype **HhDd** was mated with a non-wiry, long-haired female with the genotype **hhdd**. Complete the genetic diagram to show the ratio of offspring phenotypes expected in this cross.

<i>Parental phenotypes</i>	Wiry-haired male	Non-wiry, long-haired female
<i>Parental genotypes</i>	HhDd	hhdd
<i>Gametes</i>		
<i>Offspring genotypes</i>		
<i>Offspring phenotypes</i>		
<i>Ratio of offspring phenotypes</i>		

(3)

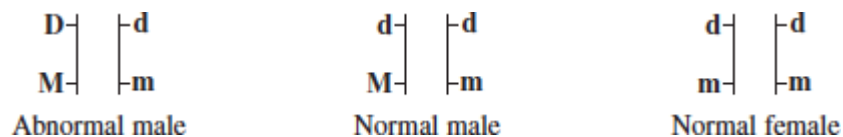
(Total 6 marks)

Q36.

- (a) Explain **one** way in which the behaviour of chromosomes during meiosis produces genetic variation in gametes.

(2)

- (b) In mosquitoes, the sex of an individual is determined by one gene. Males have the genotype **Mm** and females **mm**. Another gene is carried on the same chromosome. Normal males and females are homozygous **dd** for this gene. Abnormal males have a dominant **D** allele. The possible genotypes are shown below. The vertical lines represent homologous chromosomes.



During meiosis, allele **D** causes the homologous chromosome carrying the **m** allele to disintegrate. Cells lacking this chromosome do not develop further.

Complete the genetic diagram to show how allele **D** is transmitted from an abnormal male to his offspring.

<i>Parental phenotypes</i>	Abnormal male	Normal female
----------------------------	---------------	---------------

Parental genotypes	$\begin{array}{ c c } \hline D & d \\ \hline M & m \\ \hline \end{array}$ $\times$ $\begin{array}{ c c } \hline d & d \\ \hline m & m \\ \hline \end{array}$
Gametes	_____ _____
Offspring genotype(s)	_____
Offspring phenotype(s)	_____

(3)
(Total 5 marks)

Q37.

In a breed of cattle the **H** allele for the hornless condition is dominant to the **h** allele for the horned condition. In the same breed of cattle the two alleles **C^r** (red) and **C^w** (white) control coat colour. When red cattle were crossed with white cattle all the offspring were roan. Roan cattle have a mixture of red and white hairs.

- (a) Explain what is meant by a *dominant* allele.

(1)

- (b) Name the relationship between the two alleles that control coat colour.

(1)

- (c) Horned, roan cattle were crossed with white cattle heterozygous for the hornless condition. Complete the genetic diagram to show the ratio of offspring phenotypes you would expect.

Parental phenotypes Horned, roan × hornless, white

Parental genotypes

Gametes

Offspring genotypes

Offspring phenotypes

Ratio of offspring phenotypes

(4)

- (d) The semen of prize dairy bulls may be collected for in vitro fertilisation. The sperms in the semen can be separated so that all the calves produced are of the same sex. The two kinds of sperms differ by about 3% in DNA content.

- (i) Explain what causes the sperms of one kind to have 3% more DNA than sperms of the other kind.

(2)

- (ii) Suggest **one** reason why farmers would want the calves to be all of the same sex.

(1)

(Total 9 marks)

Q38.

Coat colour in Labrador dogs is controlled by two different genes. Each gene has a dominant and a recessive allele. The two genes are inherited independently but the effects of the alleles interact to produce three different coat colours. The table gives four genotypes and the phenotypes they produce.

Genotype	Phenotype
BbEe	black
bbEe	chocolate
Bbee	yellow
bbee	yellow

- (a) What colour coat would you expect each of the following genotypes to give?

(i) **BBEe** _____

(ii) **bbEE** _____

(2)

- (b) A **BbEe** male was crossed with a **bbee** female. Complete the genetic diagram to show the ratio of offspring you would expect.

Parental phenotypes Black male × Yellow female

Parental genotypes **BbEe** **bbee**

Gametes

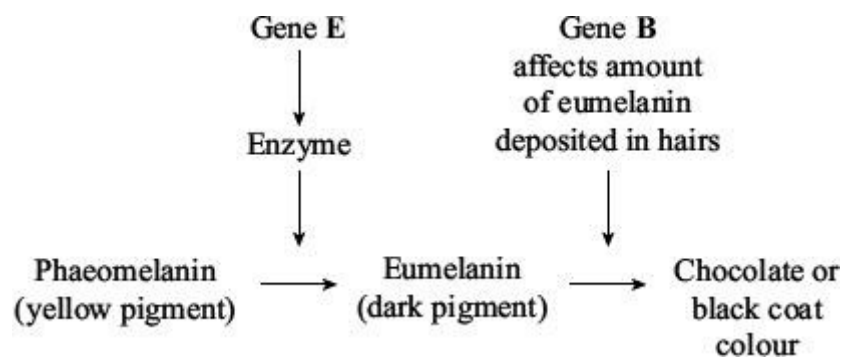
Offspring genotypes

Offspring phenotypes

Ratio of offspring phenotypes

(3)

- (c) The yellow coat colour of Labrador dogs is due to the presence of the pigment phaeomelanin in the hairs. The black and chocolate coat colours are due to different amounts of another pigment, eumelanin, deposited in these hairs. The more eumelanin there is, the darker the hair. The diagram shows the action of genes **E** and **B** in producing the different coat colours.



Use this information to explain how

- (i) the genotype **bbee** produces a yellow coat colour;

(2)

(ii) the genotype **BbEe** produces a black coat colour.

(2)

(Total 9 marks)

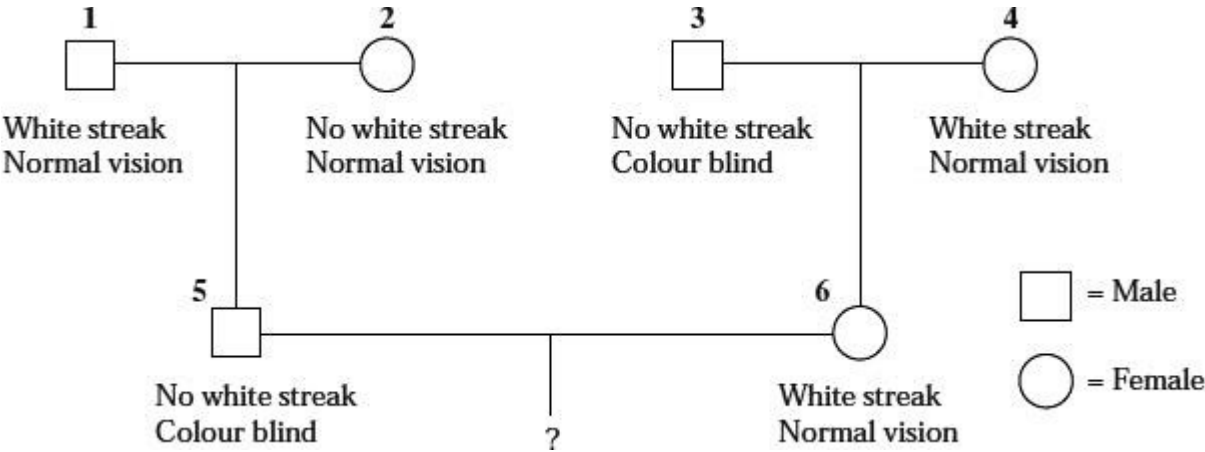
Q39.

Colour blindness is controlled by a gene on the X chromosome. The allele for colour blindness, **X^b**, is recessive to the allele for normal colour vision, **X^B**. The gene controlling the presence of a white streak in the hair is not sex linked, with the allele for the presence of a white streak, **H**, being dominant to the allele for the absence of a white streak, **h**.

(a) Explain why colour blindness is more common in men than in women.

(2)

(b) The diagram shows a family tree in which some of the individuals have colour blindness or have a white streak present in the hair.



(i) What are the genotypes of individuals **5** and **6**?

Individual **5**

Individual **6**

(2)

- (ii) Give the possible genotypes of the gametes produced by individual **5**;

individual **6**.

(1)

- (iii) What is the probability that the first child of individuals **5** and **6** will be a colour blind boy with a white streak in his hair? Show your working.

Answer _____

(2)

(Total 7 marks)

Q40.

The production of pigment in rabbit fur is controlled by two genes.

One gene controls whether any pigment is made. This gene has three alleles. Allele **A** codes for the production of one form of the enzyme tyrosinase, which converts tyrosine into a black pigment. Allele **A^h** codes for the production of a second form of the enzyme, which becomes inactive at temperatures close to a rabbit's core body temperature, so only the face, ears, legs and tail are pigmented. A third allele, **a**, fails to code for a functional tyrosinase.

The other gene controls the density of pigment in the fur. This gene has two alleles. Allele **B** is dominant and results in the production of large amounts of pigment, making the fur black.

Allele **b** results in less pigment, so the fur appears brown.

- (a) How do multiple alleles of a gene arise?

(2)

(b) The table shows some genotypes and phenotypes.

Genotype	Phenotype
A-B-	all fur black
aaB-	all fur white (albino)
A^habb	white body fur with brown face, ears, legs and tail (Himalayan)

(i) What do the dashes represent in the genotype of the black rabbit?

(1)

(ii) Give all the possible genotypes for a Himalayan rabbit with black face, ears, legs and tail.

(2)

(iii) Suggest an explanation for the pigment being present only in the tail, ears, face and legs of a Himalayan rabbit.

(2)

(c) Using the information given, explain why the phenotypes of rabbits with **AABB** and **AA^hBB** genotypes are the same.

(2)

(Total 9 marks)

Q41.

A woman comes from a family with a history of the sex-linked condition haemophilia. A

test was carried out to discover the sex of one of the embryos produced by IVF.

- (i) Explain how observation of the chromosomes from an embryo cell could enable the sex to be determined.

(2)

- (ii) The mother is known to carry the haemophilia allele. The father does not have haemophilia. What is the probability of their first child having haemophilia? Explain your answer.

(3)

(Total 5 marks)

Q42.

Coat colour in mice is controlled by two genes, each with two alleles. The genes are on different chromosomes.

One gene controls the pigment colour. The presence of allele **A** results in a yellow and black banding pattern on individual hairs, producing an overall grey appearance called agouti. Mice with the genotype **aa** do not make the yellow pigment and are, therefore, black.

The other gene determines whether any pigment is produced. The allele **D** is required for development of coat colour. Mice with the genotype **dd** produce no pigment and are called albino.

- (a) What type of gene interaction is occurring between the two genes? Explain your answer.

(2)

- (b) Give all the possible genotypes for a black mouse.

_____ (1)

- (c) An agouti mouse of unknown genotype was crossed with an albino mouse of unknown genotype. Their offspring included albino, agouti and black mice.

- (i) What was the genotype of the agouti parent?

_____ (1)

- (ii) Give **two** possible genotypes for the albino parent.

_____ (1)

- (iii) Suggest how the actual genotype of the albino parent could be determined.

_____ (2)

(Total 7 marks)

Q43.

- (a) A protein found on red blood cells, called antigen G, is coded for by a dominant allele of a gene found on the X chromosome. There is no corresponding gene on the Y chromosome.

The members of one family were tested for the presence of antigen G in the blood. The antigen was found in the daughter, her father and her father's mother, as shown in the genetic diagram below. No other members had the antigen.

	Grandmother (has antigen G)	Grandfather	Grandmother	Grandfather
<i>Genotypes</i>	_____ or _____	_____	_____	_____
<i>Gamete genotype</i>	_____ or _____	_____	_____	_____
	Father (has antigen G)		Mother	
<i>Genotypes</i>	_____		_____	

Gamete
genotypes

Daughter
(has antigen G)

Genotype

- (i) One of the grandmothers has two possible genotypes. Write these on the genetic diagram, using the symbol X^G to show the presence of the allele for antigen G on the X chromosome, and X^g for its absence.

(1)

- (ii) Complete the rest of the diagram.

(3)

- (iii) The mother and father have a son. What is the probability of this son inheriting antigen G? Explain your answer.

Probability _____

(2)

- (b) During meiosis, when the X and Y chromosomes pair up, they do not form a typical bivalent as do other chromosomes. Explain why.

(2)

(Total 8 marks)

Q44.

Figure 1 and **Figure 2** show the chromosomes from a single cell at different stages of meiosis.

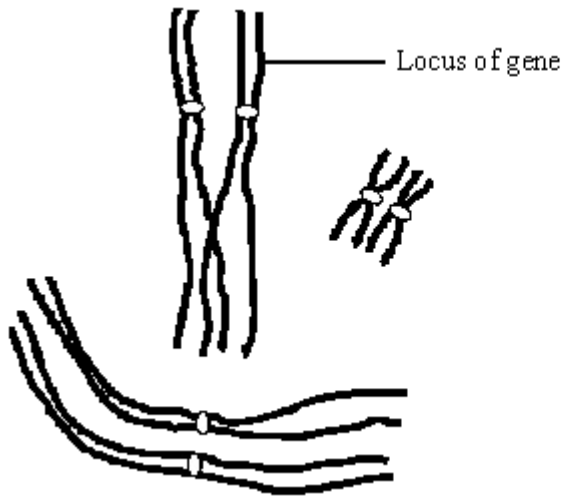


Figure 1

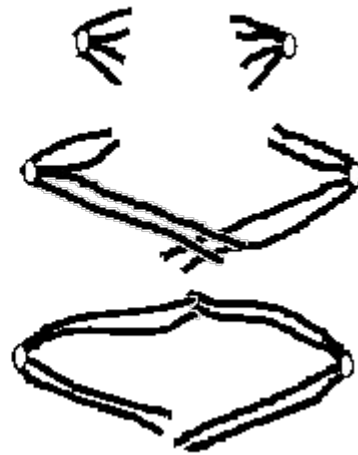


Figure 2

- (a) What is the diploid number of chromosomes in the organism from which this cell was taken?

_____ (1)

- (b) Describe what is happening to the chromosomes at the stage shown in

- (i) **Figure 1;**

(2)

- (ii) **Figure 2.**

(2)

- (c) (i) The genotype of this organism is **Bb**. The locus of this pair of alleles is shown in **Figure 1**.

Label **two** chromosomes on **Figure 2** to show the location of the **B** allele and the location of the **b** allele.

- (ii) How many genetically different gametes can be produced by meiosis from a cell with the genotype, **Bb Cc Dd**? Assume these genes are located on

(1)

different pairs of homologous chromosomes. Show your working.

(2)

(Total 8 marks)

Q45.

A sex-linked gene controls fur colour in cats. Ginger-coloured fur is controlled by the allele **G**, and black-coloured fur is controlled by the allele **g**. Some female cats have ginger and black patches of fur. They are described as tortoiseshell. Male cats cannot be tortoiseshell.

- (a) What is meant by a *sex-linked* gene?

(1)

- (b) A male cat with the genotype **X^g Y** mates with a tortoiseshell female.

- (i) Give the phenotype of the male.

(1)

- (ii) Give the genotype of the tortoiseshell female.

(1)

- (iii) Complete the genetic diagram to show the genotypes and the ratio of phenotypes expected in the offspring of this cross.

<i>Parents</i>	Male	Tortoiseshell female
----------------	------	----------------------

<i>Parental genotypes</i>	X^g Y	_____
---------------------------	------------------------	-------

Parental gametes

Offspring genotypes

Offspring phenotypes

Ratio

(3)

- (c) The effect of the **G** and **g** alleles is modified by another gene. This gene is not sex-linked and it has two alleles. The allele **d** changes the ginger colour to cream and the black colour to grey. The dominant allele **D** does not modify the effect of **G** or **g**.

A cream-coloured male cat mated with a black female whose genotype was **X^gX^g Dd**. Male kittens of two different colours were produced. Complete the genetic diagram.

*Parental
phenotypes*

Cream-coloured
male

Black
female

*Parental
genotypes*

X^gX^g Dd

*Parental
gametes*

*Male kitten
genotypes*

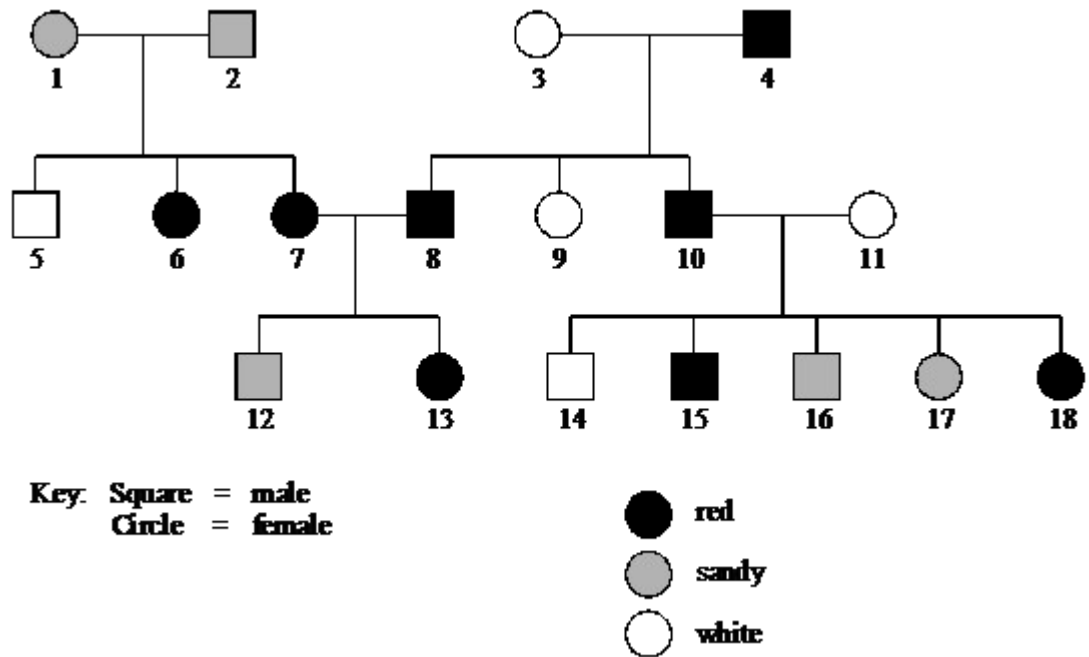
*Male kitten
colours*

(3)

(Total 9 marks)

Q46.

The diagram shows the inheritance of coat colour in pigs through three generations.



- (a) Explain **one** piece of evidence from the diagram which shows that coat colour is **not** controlled by one gene with two codominant alleles.

(3)

Two hypotheses were put forward to explain the results, each based on the action of two pairs of alleles.

	Hypothesis 1	Hypothesis 2
Phenotype	Genotype	Genotype
Red	A_B_	A_B_ or A_bb
Sandy	A_bb or aaB	aaB_
White	aabb	aabb

(_ represents either a dominant or a recessive allele of the gene)

- (b) Assuming that Hypothesis 1 is correct, give **one** possible genotype for each of the following individuals in the diagram.

11 _____

10 _____

2 _____

(2)

- (c) Explain **one** piece of evidence from the diagram which shows that Hypothesis 2 should be rejected.

(2)

- (d) Individual 18 was crossed with a pig of genotype **Aabb**.
Use Hypothesis 1 to predict the genotypes and the ratio of phenotypes expected in the offspring of this cross.

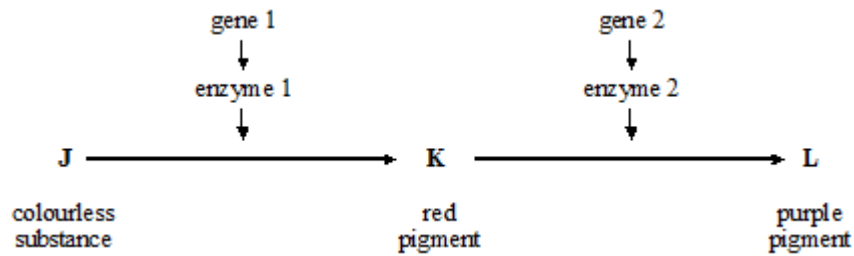
	Individual 18	Other parent
<i>Parental genotypes</i>	_____	Aabb
<i>Parental gametes</i>		
<i>Offspring genotypes</i>		
<i>Offspring phenotypes</i>		
<i>Expected ratio of offspring phenotypes</i>		

(4)

(Total 11 marks)

Q47.

A species of flowering plant can have white, red or purple flowers. The colour of the flowers is controlled by two genes. Each gene is found on a different chromosome, and is responsible for one step in a biosynthetic pathway. The biosynthetic pathway is



Gene 1 has the dominant allele **A** and the recessive allele **a**. Gene 2 has the dominant allele **B** and the recessive allele **b**. In both cases, the dominant allele needs to be present for the production of the associated enzyme.

- (a) Explain how the two genes are involved in producing white, red or purple flowers.

(6)

- (b) (i) A homozygous red-flowered plant was crossed with a homozygous white-flowered plant. All the flowers of the offspring were purple. What was the genotype of

the red-flowered parent;

the white-flowered parent?

(2)

- (ii) The purple-flowered offspring were crossed. What phenotypic ratio would you expect in the next generation? Use a genetic diagram to explain your answer.

(4)

- (c) (i) Genetically, there are different types of white-flowered plants of this species. Give their different genotypes.

(1)

- (ii) You have samples of fresh petals from the two homozygous types of white flowers, and a pure sample of the red pigment, **K**. Explain, in outline, how you might distinguish the two types of petal from each other.

(2)

(Total 15 marks)

Mark schemes

Q1.

- (a)
1. Small sample size;
 2. Fusion/fertilisation of gametes is random;
Ignore breeding is random
 3. Linked Genes;
Accept crossing over / sex linkage
 4. Epistasis;
 5. Lethal genotypes;

2 max

- (b) ttmm;

Accept mmtt or any order of these alleles e.g. mtmt, tmtm etc

1

- (c)
1. Genes are linked;
Accept 'Alleles are linked' but reject if context suggests alleles of the 'same gene'
 2. Produces few(er) tall, mottled and dwarf, normal offspring;
Accept produces few Tm and tM gametes
Accept 'fewer recombinants'
 3. Crossing over (has occurred);

3

- (d) One mark for each correct column;;

Phenotype of offspring	Ratio of offspring
Tall (plant and) normal (leaves)	9
Tall (plant and) mottled (leaves)	3
Dwarf (plant and) normal (leaves)	3
Dwarf (plant and) mottled (leaves)	1

Accept correct phenotypes in any order for one mark and correct ratio in any order e.g. 3:9:3:1 for one mark

However, phenotypes and ratio must match for two marks

Accept alternative wording e.g. short for dwarf

Accept $\frac{9}{16}$ $\frac{3}{16}$ $\frac{3}{16}$ $\frac{1}{16}$

2

[8]

Q2.

- (a) 1. LP due to mutation
OR
 Allele due to mutation;
Reject mutation caused by drinking milk.
Reject (LP) gene
2. Milk provides named nutrient;
Accept any correct named nutrient e.g. glucose, galactose, protein
Ignore 'sugar' 'lactose' as named nutrient
3. Individuals with LP more likely to survive **and** reproduce
OR
 Individuals with advantageous allele more likely to survive **and** reproduce;
Reject (LP) gene
Accept 'individuals who produce lactase' for 'LP individuals'
Accept 'pass on allele/LP/characteristic' for reproduce.
4. Directional selection;
5. Frequency of allele increases (in the offspring/next generation);
*Accept description of increasing frequency of allele e.g. 'higher proportion', 'more common' but ignore increase in **number** of allele*

4 max

- (b) 1. Dominant allele;
2. (Always) expressed/shown (when present in phenotype/offspring)
OR
 Expressed when only one (dominant allele) present;

2

- (c) 1. Mutation in promoter (DNA/gene) for transcription factor
OR
 Mutation in promoter (region/DNA) for the gene
OR
 Mutation in gene for transcription factor;
Accept mutation in an epistatic gene
2. Lactase gene continues to be transcribed/active;

2

[8]**Q3.**

- (a) 1. (Expression / appearance / characteristic due to) genetic constitution / genotype / allele(s);
2. (Expression / appearance / characteristic due to) environment;
1. *Accept: named characteristic.*
1. *Accept: homozygous / heterozygous / genes / DNA.*
1. *Ignore: chromosomes.*

2

- (b) Epistasis
OR
 Epistatic (interaction / control);
Accept: phonetic spellings.
Ignore: preceding word e.g. (recessive / dominant) epistasis. 1
- (c) **AAbb** – white
aaBB – yellow;
Both correct for one mark. 1
- (d) 1. AaBb, Aabb, aaBb, aabb;
 2. White, (white), yellow, green;
 3. 2 : 1 : 1;
Note: If genotypes are incorrect = zero marks.
 1. *Accept: equivalent genotypes e.g. ABab for AaBb.*
Accept: sequence of phenotypes does not need to mirror genotypes but must be correct.
 3. *Accept: ratios of 2:1:1 or 1:2:1 or 1:1:2 even if sequence of phenotypes do not match if mark points 1 and 2 have been awarded.*
 3. *Accept: alternative ratios in correct proportions e.g. 4:2:2*
 3. *Ignore: percentages / fractions.* 3
- (e) 1. Correct answer of 32% = **2 marks**;
 2. Incorrect answer but shows understanding that 2pq = heterozygous / carriers = 1 mark;
Accept: understanding of 2pq by using a calculation involving 2 × two different numbers. 2

[9]

Q4.

- (a) 1. Bb / suitable equivalent;
Reject sex linkage or superscripts
 2. Both parents have bar eyes, but have some offspring with round eyes, so parents must be carriers of recessive allele for round eyes; 2
- (b) 3:1; 1
- (c) Fertilisation is random
OR
 Fusion of gametes is random;
 2. Small / not large population / sample;
 3. Selection advantage / disadvantage / lethal alleles; 2 max

- (d) χ^2 / chi squared; 1
- (e) Both alleles expressed in the phenotype (if both are present); 1
- (f) 0.25; 1
- (g) 304; 1
- Award 1 mark for answers which show understanding that 2pq represents heterozygous*
- 2
- [10]**

Q5.

- (a) (Genes / loci) on same chromosome. 1
- (b) 1. GN and gn linked;
2. GgNn individual produces mainly GN and gn gametes;
3. Crossing over produces some / few Gn and gN gametes;
4. So few(er) Ggnn and ggNn individuals. 4
- (c) (Grey long:grey short:black long:black short) =1:1:1:1 1
- (d) 1. Chi squared test;
2. Categorical data. 2
- [8]**

Q6.

- (a) 1. Reduction in ATP production by aerobic respiration;
2. Less force generated because fewer actin and myosin interactions in muscle;
3. Fatigue caused by lactate from anaerobic respiration. 3
- (b) Couple **A**,
1. Mutation in mitochondrial DNA / DNA of mitochondrion affected;
2. All children got affected mitochondria from mother;
3. (Probably mutation) during formation of mother's ovary / eggs;
- Couple **B**,
4. Mutation in nuclear gene / DNA in nucleus affected;
5. Parents heterozygous;
6. Expect 1 in 4 homozygous affected. 4 max
- (c) 1. Change to tRNA leads to wrong amino acid being incorporated into protein;
2. Tertiary structure (of protein) changed;
3. Protein required for oxidative phosphorylation / the Krebs cycle, so less /

no ATP made.

3

- (d) 1. Mitochondria / aerobic respiration not producing much / any ATP;
2. (With MD) increased use of ATP supplied by increase in anaerobic respiration;
3. More lactate produced and leaves muscle by (facilitated) diffusion.

3

- (e) 1. Enough DNA using PCR;
2. Compare DNA sequence with 'normal' DNA.

2

[15]

Q7.

- (a) 1. (Expression/appearance/characteristic due to) genetic constitution/genotype/allele(s);
Accept: named characteristic.
Accept: homozygous/ heterozygous/genes/DNA.
Ignore: chromosomes.
2. (Expression/appearance/characteristic due to) environment;

2

- (b) (i) 1. (Individual) 2 has colour vision but 4 is colour blind / 10 has colour vision but 12 is colour blind
OR
4/12 is colour blind but parents have colour vision;
2. So 2/10 must be heterozygous/carriers;
Accept: (1), 2 and 4 or 10, (11) and 12.
Accept: any suitable description and explanation equivalent to points 1 and 2.
Reject: (both) parents heterozygous/carriers.
Accept: correct genotypes for 2 and 10.
Accept: for 2 marks, if it was dominant the daughters (8 and 10) of individual 4 would be colour blind.

2

- (ii) $X^B X^b$ or $X^b X^B$;
Reject: Bb / bB
Accept: $X^B X^b$ or $X^b X^B$;
Accept: use of other letter than B
e.g. $X^R X^r$, $X^H X^h$.

1

- (c) (i) 2 marks for the correct answer of 0.0625 / 6.25% / $\frac{1}{16}$;
1 mark for incorrect answer but shows 0.03125 / 3.125% / $\frac{1}{32}$;

Accept: 0.063 / 0.06 / 6.3% / 6% for 2 marks.

Accept: incorrect answer but shows / 0.0313 / 0.031 / 0.03 / 3.13% / 3.1% / 3% / $\frac{1}{4} \times \frac{1}{4}$ / 0.25×0.25 for 1 mark.

Note: if probability is calculated as a percentage but no % shown in the answer then deduct one mark. For example 6.25 = one mark, 3.125 = zero.

2

- (ii) 2 marks for the correct answer of 48(%);;
 1 mark for an incorrect answer but shows understanding that
 2pq = heterozygous or attempts to calculate 2pq;
1 mark maximum for the answer of 0.48.

2

[9]

Q8.

- (a) 1. (Reaction with ATP) breaks/allows binding of myosin to actin/ actinomyosin bridge;
 2. Provides energy to move myosin head;
1. Credit 'breaks' or 'allows' binding to actin (because cyclical)
2. Allow in context of 'power stroke' or 're-cocking' (because cyclical)
2. Ignore contraction on its own

2

- (b) (i) Any value between 68.5 and 69.49 (%);;
 If get difference of 0.9 but calculation of percentage incorrect, then award 1 mark;

2

- (ii) (Mutant mice)
 1. Unable to make phosphocreatine/ less phosphate available to make/recycle ATP;
 2. So less energy/so less ATP available for contraction/fast muscle fibres;
1 and 2. Reject production/creation of energy once
2. Accept less energy for grip
2. Accept no energy/no ATP for contraction/fast muscle fibres

2

- (c) 1. (Heterozygous) have one dominant/normal allele (for creatine production);
 2. (This) leads to production of enough/normal amount of creatine;
1. Accept has one allele/one copy of the gene for/that is making creatine

2

[8]

Q9.

- (a) Both alleles are expressed / shown (in the phenotype).
Accept: both alleles contribute (to the phenotype)
Neutral: both alleles are dominant
- (b) Only possess one allele / Y chromosome does not carry allele / gene / can't be heterozygous.
Accept: only possess one gene (for condition)
Neutral: only 1 X chromosome (unqualified)

1

- (c) 1. $X^G X^B$, $X^B X^B$, $X^G Y$, $X^B Y$;
Accept: equivalent genotypes where the Y chromosome is shown as a dash e.g. $X^G -$, or is omitted e.g. X^G
Reject: GB, BB, GY, BY as this contravenes the rubric
2. Tortoiseshell female, black female, ginger male, black male;
3. (Ratio) 1:1:1:1
2 and 3. Award one mark for following phenotypes tortoiseshell, black, (black) ginger in any order with ratio of 1:2:1 in any order.
*Allow one mark for answers in which mark points 1, 2 and 3 are not awarded but show parents with correct genotypes i.e. $X^G X^B$ and $X^B Y$ **or gametes as** X^G , X^B and X^B , Y*
3. Neutral: percentages and fractions
3. Accept: equivalent ratios e.g. for 1:1:1:1 allow 0.25 : 0.25 : 0.25 : 0.25

3

- (d) (i) Correct answer of 0.9 = 2 marks;
 Incorrect answer but shows $q^2 = 0.81$ = one mark.
Note: 0.9% = one mark
- (ii) Homozygous dominant increases and homozygous recessive decreases.

2

1

[8]

Q10.

- (a) 1. Cut (DNA) at same (base) sequence / (recognition) sequence;
Accept: cut DNA at same place
2. (So) get (fragments with gene) **R** / required gene.
Accept: 'allele' for 'gene' / same gene
- (b) 1. Each has / they have a specific base sequence;
 2. That is complementary (to allele r or R).
Accept description of 'complementary'
- (c) 1. Fragments L from parent rr, because all longer fragments / 195 base pair fragments;
Ignore: references to fragments that move further / less, require identification of longer / shorter or 195 / 135
Accept: (homozygous) recessive
2. Fragments N from parent RR, because all shorter fragments / 135 base pair fragments;
1 and 2 Accept: A3 for 195 and A4 for 135

2

2

2. *Accept: (homozygous) dominant*

3. (M from) offspring heterozygous / Rr / have both 195 and 135 base pair fragments.

Accept: have both bands / strips

Reject: primer longer / shorter

3

- (d) 1. (Cells in mitosis) chromosomes visible;
2. (So) can see which chromosome DNA probe attached to.

2

- (e) (i) 1. For comparison with resistant flies / other (two) experiments / groups;
Ignore: compare results / data / no other factors
2. To see death rate (in non-resistant) / to see effect of insecticide in non-resistant / normal flies.
Accept: 'pesticide' as 'insecticide'
Accept to see that insecticide worked / to see effect of enzyme

2

- (ii) (PM must be involved because)
1. Few resistant flies die (without inhibitor);
2. More inhibited flies die than resistant flies;
3. (PM) inhibited flies die faster (than resistant flies);
(Other factors must be involved because)
4. Some resistant flies die;
5. But (with inhibitor) still have greater resistance / die slower than non-resistant flies.
Accept: (with inhibitor) die slower than non-resistant flies

4 max

[15]

Q11.

- (a) (Recessive) allele is always expressed in females / females have one (recessive) allele / males need two recessive alleles / males need to be homozygous recessive / males could have dominant and recessive alleles / be heterozygous / carriers;

Accept: Y chromosome does not carry a dominant allele.

Other answers must be in context of allele not chromosome or gene.

1

- (b) (i) 1. 1, (2) and 5;
Accept: for 1 mark that 1 and 2 have slow (feather production) but produce one offspring with rapid (feather production).
Neutral: any reference to 3 being offspring of 1.
2. 1 must possess / pass on the recessive allele / 1 must be a carrier / heterozygous / if slow (feather production) is recessive all offspring of (1 and 2) would be slow (feather production) / if rapid (feather production) was dominant 1 would have rapid (feather

production);

Reject: both parents must be carriers / possess the recessive allele.

Reject: one of the parents (i.e. not specified) must be a carrier / heterozygous.

2

(ii) $5 = X^fY / X^fY^- / f / f^- / fY ;$

$7 = X^FX^f \text{ and } X^FX^F \text{ (either way round) /}$

or $X^FX^F \text{ and } X^FX^F \text{ (either way round) /}$

or $X^FX^f, X^fX^F \text{ and } X^FX^F \text{ (in any order);}$

Note: allow 5 = X^fY, X^fY^- .

Accept: for both 5 and 7 a different letter than F. However, lower case and capital letter must correspond to that shown in the answer. For example accept 7 = X^RX^- and X^RX^R .

2

(iii) $X^FX^f \text{ and } X^fY \text{ or } X^fX^F \text{ and } X^fY$

or $X^FX^f \text{ and } X^fY^- \text{ or } X^fX^F \text{ and } X^fY^- /$

or $Ff \text{ and } fY /$

or $Ff \text{ and } fY^- /$

or $Ff \text{ and } f^- /$

or $Ff \text{ and } f;$

Accept: a different letter than F. However, lower case and capital letter must correspond to that shown in the answer.

Accept: each alternative either way round.

1

(c) Correct answer of 32 (%) = 3 marks;;;

Accept: 0.32 = 2 marks

If incorrect answer, allow following points

1. $p^2 / q^2 = 4\% / 0.04 / \text{or } p / q = 0.2;$

2. Shows understanding that $2pq = \text{heterozygotes / carriers};$

Accept: answer provided attempts to calculate $2pq$. This can be shown mathematically i.e. 2 x two different numbers.

3

[9]

Q12.

(a) 1. Expression / appearance / characteristic due to genetic constitution / genotype / allele(s);

1. Accept: named characteristic

1. Accept: homozygous / heterozygous / genes / DNA

1. Neutral: chromosomes

2. (Expression / appearance / characteristic) due to environment;

2

- (b) (i) 1. 3 and 4 and 9 / 11 / affected offspring;
1. Accept: 9 / 11 and their parents
1. Accept: unaffected parents have affected children
2. Both 3 and 4 are carriers / heterozygous;
2. Accept: if 3 and 4 are unaffected all their children will be unaffected

OR

If dominant at least one of 3 and 4 would be affected;

2

- (ii) 1. 11 is affected, 3 is not;
1 Accept: 3 / unaffected father / parents produce an affected daughter
1. Accept: 3 and 4 would only produce unaffected females
2. 3 / father of 11 does not have a recessive allele on his X chromosome / X^t ;
2. Answers must be in context of alleles

OR

(If on X) 11 / affected female would not receive the recessive allele on X chromosome / X^t from 3 / father;

Reject: recessive / dominant chromosomes

OR

(If on X) 3 / father (of 11) would pass on the dominant allele on his X chromosome / X^T ;

2

- (c) (i) Answer in range of $5.8 - 6.2\% = 3$ marks;;;
Answers in range of $0.058 - 0.062 = 2$ marks

If incorrect answer, then 2 max of following points

1. $q^2 / p^2 / tt = 0.001$ or 1 divided by 1000;
2. $p / q / T = 0.968 - 0.97$;
3. Understanding that heterozygous = $2pq$;
3. This can be shown mathematically ie $2 \times$ two different numbers
3. Accept: answer provided attempts to calculate $2pq$

3 max

- (ii) Affected individuals (usually) do not reproduce / die during childhood / do not pass on allele / genetic screening;

1

[10]

Q13.

- (a)
- 2.84:1
- ;

Accept '2.84 to 1' or (just) 2.84
Do not accept 1:2.84 or 142:50

1

- (b) 1. Some embarrassed / some not willing to show tongue / cannot tell;
 2. Could not decide whether thumb was straight or not / thumb bending is judgemental / subjective;

2

- (c) 1. (No) - should be 92.9% / should be calculated from 182 out of 196 / should not be calculated from 182 out of 200;

Allow either no or yes approach but no mark awarded for no or yes on its own

2. (Yes) – assumes 4 out of 200 use either hand;

Accept ambidextrous

3. (But) sample may not be representative;

This could be expressed in other ways e.g. only based on one part of the country / might not be the same in different parts of the UK / might not be representative of UK

4. Small sample size / only sampled 200;

2 max

[5]**Q14.**

- (a) (i) 1. No overall pattern / pattern (of right or left most common) is not the same for all islands;
Allow expression in other ways e.g. three islands show left on top is more common
2. For **(B) C** and **E** there is little difference;
3. Large differences on **A** and **D** and opposite ways (to each other);
Need both aspects but allow other expressions of 'opposite ways'

2 max

- (ii) 1. Can record all individuals on (small) islands;
2. (So) no / less sampling error;
3. (Maybe) different rates of mutation / different selection pressures / different environmental conditions;
4. Inbreeding / breeding with close relatives (more likely);
5. (Little) gene flow / (more chance of) genetic drift;
Accept reference to either of these ideas for this point

2 max

- (b) 1. If R is recessive, R × R parents cannot produce L offspring;
Accept use of genetic diagrams to illustrate points 1 and 2
2. If L is recessive, L × L parents cannot produce R offspring;
Accept right arm on top as R etc.
3. R × R **and** L × L parents produce both types of offspring;
Need reference to two parent crosses for this mark

3

- (c) Both L and R in a set of twins / (some) twins show different arm-folding;

1

[8]

Q15.

- (a) 1. Large number of eggs / offspring / flies (therefore) improves reliability / can use statistical tests / are representative / large sample (size) / reduces sampling error;

*Each mark point requires a feature linked in mark scheme
(by therefore) to an explanation*

*Do not accept a large number of eggs produces a large
number of flies unless the term sample is used*

Ignore references to accuracy or precision

2. Small size / (breed) in small flasks / simple nutrient medium (therefore) reduces costs / easily kept / stored;

Accept small size so can be kept in small flasks

3. Size / markings / phenotypes (therefore) males / females easy to identify;

*Answers must relate to size, markings or use the term
phenotype*

4. Short generation time / 7 - 14 days / develop quickly / reproduce quickly (therefore) results obtained quickly / saves times / many generations;

2 max

- (b) (i) 1. $X^R X^R$ and $X^r Y$;
*All marking points are completely independent. Allow
crosses from the following parents for a possible three
marks:*

$X^R X^R$ and X^r -

$X^R X^R$ and $X^r Y$;

RR and rY / rY^-

RR and $r-$ or RR and r

2. X^R and X^R plus X^r and Y ;

3. $X^R X^r$ and $X^R Y$;

OR

1. $X^R X^r$ and $X^r Y$;

OR

$X^R X^r$ and $X^r -$

$X^R X^r$ and $X^r Y$;

2. X^R and X^r plus X^r and Y ;
 Rr and rY / rY^-
 Rr and r^- or Rr and r
Accept different symbols e.g. W and w
2. *Accept gametes in a punnet square*
3. $X^R X^r$ and $X^r Y$;

3

- (ii) Fertilisation is random / fusion of gametes is random / small / not large
population / sample / selection advantage / disadvantage / lethal alleles;
Mutation = neutral
Random mating = neutral
Accept fertilisation / fusion of gametes is due to chance

1

- (c) 1. Males have one allele;
Answers should be in context of alleles rather than chromosomes
2. Females need two recessive alleles / must be homozygous recessive /
could have dominant and recessive alleles / could be heterozygous /
carriers;

2

[8]

Q16.

- (a) Is always expressed / shown (in the phenotype);
Reject 'is always present' without further qualification

1

- (b) $C^B C^B$, $C^B C^P$ and $C^B C^Y$;
All three are required for the mark

Or

$C^B C^B$, $C^P C^B$ and $C^Y C^B$;
*Accept $C^B C^B$, $C^B C^P$, $C^B C^Y$,
 $C^Y C^B$ and $C^P C^B$*
*Accept BB , BP and BY or
 BB , BP , BY , YB and PB*

1

- (c) 1. Two genotypes (as parents) shown as $C^P C^Y$
*Award **one mark maximum** for candidates who have
misread the question and complete a correct genetic cross
between a pink snail, $C^P C^Y$ and a yellow snail, $C^Y C^Y$ to give
pink and yellow offspring*

Or

Two sets of gametes shown as C^P and C^Y ;

2. Genotypes of offspring shown as $C^P C^Y$, $C^P C^P$ and $C^Y C^Y$;
3. Above genotypes of offspring correctly linked to phenotypes i.e. pink and yellow;

Accept ratio (or equivalent) of 3 pink: 1 yellow for mark point 3

3

- (d) 1. Correct answer of 42% = 3 marks
Answer of 0.42 = 2 marks
*Award **one mark maximum** for answer of 49.9 / 49.98 / 50% or 0.49 / 0.5*
2. $q^2 = 0.49 / 49\%$ **OR** $q = 0.7 / 70\%$
*Award **one mark maximum** for answer of 40.8 / 41% or 0.41*
3. Shows understanding that $2pq$ = heterozygotes / carriers / shows answer is derived from $2pq$;

Accept: $b^2 = 0.49 / 49\%$ or $b = 0.7 / 70\%$ for mark point 2

3

[8]

Q17.

- (a) (i) 1. Animal 2 / 5 has hair but offspring do not;
Accept parents as alternative to animals 2 and 5
2. So 2 / 5 parents must be heterozygous / carriers;
1 + 3: Allow reference to children / offspring for animals 7 + 8

OR

3. 4 / 7 / 8 are hairless but parents have hair;
Ignore reference to individuals 1 and 6
4. So 2 / 5 must be heterozygous / carriers;

2

- (ii) Hairless males have fathers with hair / 4 is hairless but 1 is hairy / 7 and / or 8 are hairless but 6 is hairy / only males are hairless;
Ignore references to other individuals
Ignore reference to genotypes
Allow credit for candidate who states that evidence is not conclusive / pedigree possible with autosomal character;

1

- (b) 1. Parental genotypes
 $X^H X^h$ and $X^H Y$
Gametes
 $X^H X^h X^H Y$;
Accept any letter for gene but capital letter must represent

dominant allele.

Both parental genotypes and gametes must be correct

2. Genotypes of offspring
 $X^H X^H$, $X^H Y$, $X^H X^h$, $X^h Y$;
Allow for offspring genotypes correctly derived from gametes given by candidate;
3. Phenotypes of offspring
 female with hair
 male with hair
 male hairless;
Allow phenotypes correctly derived from offspring genotype
Allow $H \equiv X^H$, $h \equiv X^h$
4. 0.25 / $\frac{1}{4}$ / 1 in 4 / 25 %
Ignore 1:3 in context of correct probability
Reject 1:4

4

[7]

Q18.

- (a) (i) 1. Parents are heterozygous;
Accept carriers / carries white allele
2. Kittens receive white allele from parents / black cat;

1 max
- (ii) 1:1;
Answer must be expressed as a ratio that could be reduced to 1 : 1

1
- (b) (i) Black,
 Chocolate,
 Black;
All three correct for the mark

1
- (ii) Parental phenotypes Chocolate male Black female
 1. Parental genotypes bb^i Bb^i ;
Both genotypes needed for the mark.

1
 2. Parental gametes b b^i B b^i ;
Allow credit if gametes are correctly derived from candidate's incorrect parental genotypes.

1
 3. Offspring genotypes Bb , Bb^i bb^i $b^i b^i$;

*Genotype(s) must be with correct phenotype.
 Allow credit if symbols other than B / b / b have been used correctly.
 Ignore genetic diagrams unless clearly annotated.*

1

Offspring phenotypes Black Chocolate cinnamon;

- (iii) 1. Offspring ratios are a probability / not fixed / arise by chance /
 2. gametes may not be produced in equal numbers /
 3. fertilisation / fusion of gametes is random /
 4. small sample;

1

- (iv) 1. Possible if parents homozygous / bb;
 2. Don't know genotype of chocolate cat / chocolate cat could be homo- or heterozygous / chocolate cat could be bb or bb';
 3. Two chocolate cats could give cinnamon kittens;

2 max

[9]

Q19.

- (a) (i) Only expressed / shown (in the phenotype) when homozygous / two (alleles) are present / when no dominant allele / is not expressed when heterozygous;

1

- (ii) Both alleles are expressed / shown (in the phenotype);
Allow both alleles contribute (to the phenotype).

1

- (b) (i) Evidence (not a mark)

3 and 4 / two Rhesus positives produce Rhesus negative child / children / 7 / 9;

Explanation (not a mark)

Both Rhesus positives / 3 and 4 carry recessive (allele) / are heterozygous / if Rhesus positive was recessive, all children (of 3 and 4) would be Rhesus positive / recessive;

Do not negate mark if candidate refers to gene rather than allele.

Answers including correct and incorrect evidence = zero marks evidence and explanation.

2

- (ii) Evidence (not a mark)

3 would not be / is Rhesus positive / would be Rhesus negative;

Explanation (not a mark)

3 would receive Rhesus negative (allele) on X (chromosome) from mother / 3 could not receive Rhesus positive (allele) from mother / 3 would not receive Rhesus positive (allele) / X (chromosome) from father / 1 / 3 will receive Y (chromosome) from father / 1;

OR

Evidence (not a mark)

9 would be Rhesus positive / would not be / is Rhesus negative / 8 and 9 / all daughters of 3 and 4 would be Rhesus positive;

Explanation (not a mark)

As 9 would receive X chromosome / dominant allele from father / 3;

Do not negate mark if candidate refers to gene rather than allele.

One mark for evidence and one mark for explanation linked to this evidence.

Any reference to allele being on Y chromosome negates mark for explanation.

2

- (c) Correct answer of 48(%) = 3 marks;;;

$$q^2 / p^2 = 16\% / 0.16 / p / q = 0.4;$$

Shows that $2pq$ = heterozygotes / carriers;

Final answer of 0.48 = 2 marks

Allow mark for identifying heterozygotes if candidate multiplies incorrect p and q values by 2.

3

[9]

Q20.

- (a) Cannot make (active) enzyme A (which converts precursor to linamarin) / cannot make linamarin;

1

- (b) (i) **AL + Al + aL + al ;**

1

- (ii) Meiosis separates alleles / homologous chromosomes / pairs of chromosomes;
Independent assortment / means either of **A** / **a** can go with either of **L** / **l**;

Accept "random segregation" but cancel if reference to crossing-over

2

- (c) From parental genotypes: **AaLl** × **AaLl** (no mark)
Note: If wrong parental genotypes / wrong gametes: ALLOW correct derivation of offspring genotypes = 1 max

Correct derivation of offspring genotypes; max 2 marks if error in Punnett square

	AL	AI	aL	al
AL	AALL	AALI	AaLL	AaLI
AI	AALI	AAII	AaLI	AaII
aL	AaLL	AaLI	aaLL	aaLI
al	AaLI	AaII	aaLI	aall

Correct identification of offspring genotypes with at least one **A** and two **I** alleles (= grey cells in above table);

Correct proportion: 3 / 16 / 3:13 / 18.75% ;

3

- (d) (i) There was no (significant) difference in damage between cyanogenic and acyanogenic / being cyanogenic has no effect;

1

- (ii) The difference (from expected / from chance variation) is significant / difference / results not just due to chance;
Reject null hypothesis;
Being cyanogenic does help protect from slug damage;

3

- (e) High slug population:

- Find only cyanogenic plants / only cyanogenic plants survive;
- (Cyanide release) limits / stops feeding by slugs / slugs killed;

Accept: converse argument re. acyanogenic plants

Low slug population:

- Find both types of plant;
- Less selection pressure on plants from slugs / no selective advantage / no selection / described;

4

[15]

Q21.

- (a)
 - Homologous chromosomes pair up / bivalents form;
 - Crossing over / chiasmata form;
 - Produces new combination of **alleles**;
 - Chromosomes separate;
 - At random;
 - Produces varying combinations of chromosomes / genes / alleles (*not twice*) ;
 - Chromatids separated at meiosis II / later;

Independent assortment / random segregation = marking points 4 and 5

6 max

- (b) (i)

Parental phenotypes Agouti White

Parental genotypes BbAa bbaa ;

Gamete genotypes	BA	Ba	bA	ba	ba	;
Offspring genotypes	BbAa	Bbaa	bbAa	bbaa		;
Offspring phenotype	Agouti	Black	White	White		;

Phenotypes must match genotypes

Allow marking points 2 and 3 if correctly derived from wrong parental genotypes

4

(ii)

Colour of offspring	Observed (O)	Expected (E)	(O-E)	(O-E) ²	$\frac{(O-E)^2}{E}$
Agouti	34	30	4	16	0.53
Black	35	30	5	25	0.83
White	51	60	9	81	1.35
					$\sum \frac{(O-E)^2}{E} = 2.71 \text{ or } 2.72$

∴ 2

(χ^2 correct = 2 marks)

((O-E)² all correct = 1 mark)

p = 0.05;

2 degrees of freedom;

Differences due to chance / no significant difference as χ^2 less than / to left of critical value OR Not due to chance / difference is significant as χ^2 greater than to right of critical value;

(as appropriate for candidates χ^2)

3

[15]

Q22.

(a) Table completed as below:

Kingdom	Animalia / Animals
Phylum	Chordata
Class	Mammalia
Order	Rodentia
Family	Caviidae
Genus	Cavia

Column 1 correct;

Species	<i>porcellus</i>

Column 2 correct;

2

- (b) Mutation occurs;

Correct e.g. of isolating mechanism

e.g.

temporal – different breeding seasons / feeding times /

ecological / behavioural – different courtship displays / different niches /

habitats / feeding areas /

mechanical – mismatch of reproductive parts /

gamete incompatibility – sperm killed in female's reproductive tract /

hybrid inviability / hybrid infertility;

Ignore references to "genetic isolation" or "reproductive isolation"

Different selection pressures operate / changes in allele frequency /

divergence of gene pools;

3

- (c) Using candidate's symbols for alleles –
e.g. B = black, b = brown, S = short, s = long:

Parental genotypes correct:

Male **A**
SSBb

Female **B**
SsBB;

Gametes correctly derived from

candidate's parental genotypes: SB Sb SB sB;

offspring genotypes correctly

derived from candidate's

suggested gametes – accept Punnett square or line diagram;

offspring genotypes correct: SSBB SsBB SSBb SsBb;

If monohybrid: cross $\Rightarrow$ 0 marks

4

- (d) There is no (significant) difference between observed and expected results / any difference is due to chance;

1

[10]

Q23.

parental genotypes correct: $X^R X^r$ AND $X^R Y$;

gametes correct for candidate's parental genotypes;

offspring genotypes correct and colourblind male identified as $X^r Y$ /

correct genotypes derived from cand's gametes and identify $X^r Y$;

correct probability = $\frac{1}{4}$ / 0.25 / 25% / 1 in 4 / 1:3 ;

[4]

Q24.

- (a) (variation in) temperature will affect the solubility of oxygen / rate of respiration / use of oxygen by cells / diffusion / gas exchange;

to gain credit point made must concern oxygen

1

- (b) (i) there is no difference between the partial pressure of oxygen in the two groups / the partial pressure of oxygen is the same in each group;

1

- (ii) results may have been due to chance and statistical test allows us to determine the probability of this / of the difference between results being significant;
enables acceptance or rejection of null hypothesis;
The key points here are chance and probability used in the correct context.

2

- (c) **A**;
because partial pressure of oxygen only reduced when zinc in water / in **Y** / because when injected zinc / in **X** has no effect on partial pressure of oxygen in blood;

2

- (d) less oxygen transport to cells / in fish / in blood;
anaerobic respiration;
lactic acid produced / less carbon dioxide removed (from gills);
more H⁺;

3 max

- (e) (i) copper;
calculation based on comparing concentration in woodlice with that in leaves;
accept any suitable method here, giving marks for the method and explanation. For example, calculating ratio of concentration in woodlice to concentration in leaves.

2

- (ii) not absorbed from gut / passes out in faeces / egested / urine / excreted;

1

- (iii) woodlice eat large amount of leaves;
copper stored / accumulates in body;

2

- (f) (i) mutation;

1

- (ii) (as a component of) nucleic acids / DNA / RNA / nucleotides;
phospholipids;
ATP / ADP;

2 max

- (iii) arsenic-tolerant plants would not be able to take up phosphates / take up a little phosphate;
since likely to involve same mechanism / same carrier / protein;
(process of) growth would be poorer than non-tolerant plants;

3

[20]

Q25.

- (a) Parents genotypes Aabb aaBb ;
Gametes formed Ab ab aB ab ;

if parental genotypes wrong allow correctly derived gametes only

Offspring genotypes AaBb Aabb aaBb aabb

and

Offspring phenotypes 1 Walnut ; 1 Pea : 1 Rose : 1 single ;

*Just **one** mark for offspring genotypes **and** phenotypes*

If parents not diploid, no marks gained

3

- (b) Correct answer 0.6, however derived, scores 2 marks
Wrong answer, but evidence of correct working
(e.g. $p^2 / q^2 = 0.36$) scores 1 mark

2

[5]

Q26.

- (a) (i) Two, as white blood cells are diploid cells / alleles are present on each chromosome of an homologous pair / one maternal and one paternal;

1

- (ii) **A and B**

(reject I^A and I^B)

1

- (b) 1 in 8 / 1 / 8 / 12.5% / 1:7 / 0.125;
(Reject 1:8) parents $I^A I^O$ and $I^B I^O$;
give 1:3 / $\frac{1}{4}$ / 1 in 4 / 25% probability of blood group A and half will be male;
(accept 2nd and 3rd points from a suitable genetic diagram)

3

[5]

Q27.

- (a) (i) where a change triggers a response which reduces the effect of a change;

1

- (ii) e.g. sweating, breathing, defaecating, other valid example;

(reject respiration

evaporation not acceptable as a 2nd mark if sweating or breathing given)

2 max

- (iii) hypothalamus;

1

- (b) (i) pituitary;

(ignore anterior pituitary)

1

- (ii) 1. ADH causes vesicles containing aquaporins / aquaporins to be inserted into membrane / collecting duct wall / plasma;
2. water enters cell through aquaporins;
3. by osmosis / diffusion / down a water potential gradient;

4. (from cell) to capillary;
5. via interstitial fluid;

4 max

(c) (i) excessive urination / drinking / diluted urine / thirst;

1

(ii) because males only have one X chromosome / do not have Y chromosome;
a single copy of the recessive allele will be expressed;

2

(iii) recessive alleles can be carried by individuals without showing effects / dominant allele always expressed;
organism that are carriers more likely to reproduce / affected organism less likely to reproduce;
therefore recessive alleles are more likely to be passed on / dominant alleles less likely to be passed on;

3

[15]

Q28.

Parental genotypes: Gg nn gg Nn ;
Gamete genotypes Gn gn gN gn ;

	gN	gn
Gn	Gg Nn Grey, normal	Gg nn Grey, vestigial
gn	gg Nn Ebony, normal	gg nn Ebony, vestigial

All offspring genotypes correct;

All offspring genotypes correctly derived;

[4]

Q29.

(a) (i) Only seen in males / not in females;

1

(ii) Unaffected parents / mother → child with M.D. /
(1 ×) 2 → 5 / (3 ×) 4 → 11 / 8 (× 9) → 13;

1

(b) 5 = X^dY

6 = X^DY

$$7 = X^D X^d \text{ AND } X^D X^D$$

$$8 = X^D X^{d+};$$

All 4 correct = 2 marks

2 or 3 correct = 1 mark

max 2

- (c) $\frac{1}{4}$ / 0.25 / 25% / 1:3 / 1 in 4; (NOT '1:4')

1

[5]

Q30.

- (a) Cannot make (active) enzyme A (which converts precursor to linamarin) / cannot make linamarin;

1

- (b) (i) **AL + AI + aL + al** ;

1

- (ii) Meiosis separates alleles / homologous chromosomes / pairs of chromosomes;
Independent assortment / means either of **A** / **a** can go with either of **L** / **l**;

[Accept: 'random segregation'] [Cancel: if reference to crossing-over]

2

- (c) From parental genotypes: **AaLl** × **AaLl** (no mark)

[Note: If wrong parental genotypes / wrong gametes: ALLOW correct derivation of offspring genotypes] (= max 1)

Correct derivation of offspring genotypes:

	AL	Al	aL	al
AL	AALL	AALl	AaLL	AaLl
Al	AALl	AaLl	AaLl	Aall
aL	AaLL	AaLl	aaLL	aaLl
al	AaLl	AaLl	aaLl	aall

Correct identification of offspring genotypes with at least one **A** and two **l** alleles (= grey cells in above table);

Correct proportion: 3 / 16 / 3:13 / 18.75% ;

3

- (d) (i) There was no (significant) difference in damage between cyanogenic and acyanogenic / being cyanogenic has no effect;

1

- (ii) The difference (from expected / from chance variation) is significant / difference / results not just due to chance;
Reject null hypothesis;

Being cyanogenic does help protect from slug damage;

3

- (e) High slug population:
1. Find only cyanogenic plants / only cyanogenic plants survive;
 2. (Cyanide release) limits / stops feeding by slugs / slugs killed;
[Accept: converse argument re. acyanogenic plants]

Low slug population:

3. Find both types of plant;
4. Less selection pressure from slugs / no selective advantage / no selection / described;

4

[15]

Q31.

- (a) Gg / suitable equivalent;
Grey : black about 3: 1;

[Note: Can be in table / diagram]

2

- (b) To determine the probability;
[Accept: Likelihood]
Of the results being due to chance;
[Accept: Coincidence]

2

- (c) (i) both alleles will be expressed (in the phenotype);

1

- (ii) 0.25 / 25%; = 2 marks
 $C^N = 250 / 1000$; = 1 mark

2

- (iii) $P^2 = (0.25)^2 / 0.0625$ / square of calculated figure for C^N ; = 2 marks
 $p^2 + 2pq + q^2 = 1.0$; = 1 mark
= 31.25 / 31;
[Accept: Derived from either p^2 or q^2]

3

[10]

Q32.

- (a) Only expressed in the homozygote / not expressed in the heterozygote / not expressed if dominant present;

1

- (b) Tt Aa × tt aa ;
TA Ta tA ta ta ;

	TA	Ta	tA	ta	
ta	TtAa	Ttaa	ttAa	ttaa	;
	Orange	Orange	White	White	;

	striped	unstriped	striped	unstriped / snowy
--	---------	-----------	---------	-------------------

If parental genotype incorrect allow 1 mark for correct gametes based on given genotype and 1 mark for correct cross based on these gametes = 2 max MUST be clear link between F1 genotype and phenotype.

4

- (c) (White) not camouflaged / not got stripes / white colour stands out;

Prey can take avoidance or are aware earlier / sooner;

Must have a time reference

2 max

[7]

Q33.

- (a) (i) BBX^AY , BbX^AY ;

1

- (ii) BbX^AX^a , bbX^AX^a ;

1

- (b) *parental genotypes* – BbX^AY x BbX^AX^a ;

1

Gametes – $(BX^A, bX^A,)BY, bY,$ $BX^A, B X^a, bX^A, b X^a$;

1

Genotypes of sons- ;

		Male gametes	
		BY	bY
Female gametes	BX^A	BBX^AY	BbX^AY
	$B X^a$	$BB X^a Y$	$Bb X^a Y$
	bX^A	BbX^AY	bbX^AY
	$b X^a$	$Bb X^a Y$	$bb X^a Y$

1

0.125 / 12.5% / 1/8 ;

1

[6]

Q34.

- (a) aabb;

1

- (b) AaBb and aabb;

1

- (c) Pea comb offspring will produce blue eggs;
Alleles **A** and **B** are inherited together / are on the same chromosome;
2
- (d) Reference to crossing over;
Reduce chance of genes being separated (by crossing over);
If crossing over occurred some gametes will contain alleles **A** and **b**;
2 max
- (e) Two suitable environmental factors;
e.g.
Diet / named component of diet;
Temperature;
Light intensity / duration;
Disease;
2 max
- (f) Cross $C / X^f X^f$ and $X^F Y$;
1
- (Only) cross where all males are one phenotype and all females are a different phenotype;
Cross showing all males are slow feather production, all females fast feather production;
2
- (g) Two alleles for each gene present in male / chromosomes are homologous in male;
Female has one allele for each gene;
Recessive alleles always expressed in female;
Males need two recessive alleles for allele to be expressed / in males recessive alleles can be masked by dominant allele
3 max

[14]

Q35.

- (a) hhDD, hhDd;
(both correct 1 mark)
1
- (b) Epistasis;
One gene controlling / inhibiting the expression of another;
2
- (c) Gametes correct HD, Hd, hD, hd, hd
(correct for both parents);
- | | |
|------------|--|
| Genotypes | HhDd, Hhdd, hhDd, hhdd ; |
| Phenotypes | wiry wiry non-wiry, short non-wiry, long |
| Ratio | 2 1 1 ; |
- 3

[6]

Q36.

(a) *Two linked points:*

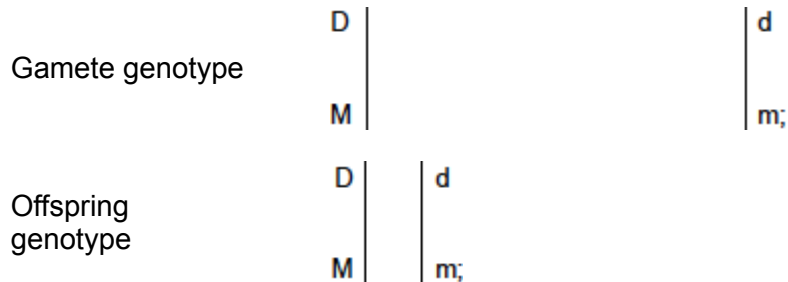
Crossing over / exchange of material (between chromatids);
Different combinations of alleles / linkage groups changed / broken;

OR

Independent assortment / alignment of (homologous) chromosomes;
Different combinations of (maternal and paternal) chromosomes / alleles;

2 max

(b)



Offspring phenotypes Abnormal males / (all) (no females);

3

[5]

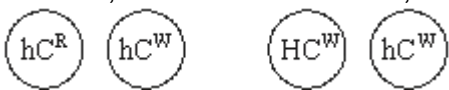
Q37.

(a) is always expressed(in the phenotype) / produces (functional) proteins;

1

(b) codominance;

1

(c) *Parental genotypes* - hhC^RC^w , HhC^wC^w ;

Gametes-
Offspring genotypes - HhC^RC^w , hhC^RC^w , HhC^wC^w , hhC^wC^w ;
Offspring phenotypes - hornless roan, horned roan, hornless white, horned white
Ratio of offspring - 1 1 1 1;

4

(d) (i) sperm(with more DNA) have X chromosome;
X is larger / has more genes than Y;

2

(ii) female for milk / males for meat / male or female for breeding;

1

[9]

Q38.

(a) (i) black;

1

- (ii) chocolate; 1
- (b) **BE, Be, bE, be** and **be**;
BbEe, Bbee, bbee, bbEe;
 1 black: 2 yellow: 1 chocolate; 3
- (c) (i) no enzyme coded for when no dominant / **E** allele;
 phaeomelanin not converted – (remains yellow); 2
- (ii) **E** allele results in enzyme producing eumelanin;
B allele - more eumelanin deposited in hairs; 2
- [9]**

Q39.

- (a) males are XY and females XX / males have one X chromosome and females two X chromosomes;
 males only have one allele (of the gene) present / recessive allele always expressed;
 colour blindness is masked in heterozygote / female needs 2 recessive alleles to be colour blind; 2 max
- (b) (i) 5 - hh X^b Y;
 6 - Hh X^B X^b; 2
- (ii) h X^b , h Y, and H X^B, h X^B, H X^b, hX^b; 1
- (iii) 1 / 8 or 12.5% or 0.125;;
- either*
 genetic diagram to show genotypes Hh X^b X^b, Hh X^BY, hh X^B X^b, hh X^BY, HHX^bX^b, Hh X^bY, hh X^b X^b; hh X^bY;
 1 / 8;
or
 P (boy) = 0.5, P (colour blind) = 0.5, P (white streak) = 0.5;
 (0.5 × 0.5 × 0.5 =) 0.125; 2
- [7]**

Q40.

- (a) mutations;
 which are different / at different positions in the gene; 2
- (b) (i) either dominant or recessive allele; 1
- (ii) A^hA^h BB, A^haBB, A^hA^h Bb, A^haBb;
 (allow 1 mark for 2 or 3 correct answers) 2

- (iii) temperature lower at extremities;
enzyme active / not denatured;

2

- (c) if allele A is present (normal) tyrosinase / enzyme is produced, so it does not matter what other allele is present / explanation of why heterozygote is same phenotype as double dominant in terms of enzyme produced;
phenotype / rabbit is black as both have alleles A and B;

2

[9]

Q41.

- (i) female XX, male XY;
Y shorter / smaller than X;
- (ii) haemophilia is a recessive allele;
defective allele (gene) present on X, missing from Y;
male 0.5(50% / $\frac{1}{2}$) probability of haemophilia;
female 0 / no chance;
(0.25(25% / $\frac{1}{4}$) first baby having haemophilia);

2

or

$X^H X^h$ $X^H Y$;
 $X^H X^H + X^H X^h + X^H Y + X^h Y$;
 $X^h Y$ is a sufferer

3 max

[5]

Q42.

- (a) epistasis;
one gene influences the expression of another / description using example in question;
- (b) aaDD, aa Dd (or DDaa, Ddaa);
- (c) (i) AaDd (or DdAa);
- (ii) aadd, Aadd (or ddaa, ddAa);
- (iii) cross with black individual / genotype aaDd or aaDD;
genotype is Aadd if agouti offspring / genotype is aadd if no agouti offspring;
Accept;
repeat cross using original parents many times;
ratio is 4 albino : 3 agouti : 1 black if Aa, or 2 albino : 1 agouti : 1 black if aa;

2

1

1

1

2

[7]

Q43.

- (a) (i) paternal grandmother: $X^G X^G$ or $X^G X^g$ 1
- (ii) grandparent genotypes: $[X^g Y]$ $[X^g X^g]$ $[X^g Y]$;
 gametes: $[X^G$ and X^g , or X^G only] $[X^g$ and $Y]$ $[X^g]$ $[X^g$ and $Y]$;
 parents genotypes: $[X^G Y]$ $[X^g X^g]$
 gametes: $[X^G$ and $Y]$ $[X^g]$
 daughter: $[X^G X^g]$;
(all correct = 3 marks);
(max 2 if no distinction between pairs of gamete genotypes,
e.g. comma, space or circle);
(allow omission of gametes clearly not involved in next
generation);
(all males XY and females XX = 1 mark, if no other marks); 3
- (iii) nil;
 X chromosome, without **G** allele, inherited from mother / Y must
 be inherited from father, not X^G ; 2
- (b) X and Y chromosomes are different sizes / shapes;
 chromatids unable to line up and form bivalent / only
 short pairing region / most of length not homologous; 2

[8]**Q44.**

- (a) 6; 1
- (i) chromosomes are arranged in (homologous) pairs / bivalents;
 crossing over / chiasma present / exchange of genetic information;
 bivalents arranged independently; 2 max
- (ii) separation / splitting / pulling apart of homologous chromosomes /
pairs of chromosomes;
(must give indication that one chromosome moves to each
side)
(must be in the context of meiosis – not chromatid
movements and not chromosomes separate)
 pulled at centromere / by spindle / fibres; 2
- (c) (i) the short arm of both chromosomes labelled on the middle
 homologous pair;
*(**B** and **b** must be labelled on separate chromosomes)* 1
- (ii) 8 = 2 marks;
 working showing genotypes with 1 allele from each pair
 (for example, **B C D**) = 1 mark 2

Q45.

- (a) gene located on X / Y / one sex chromosome;
(allow gene on X or Y chromosome, not X and Y) 1
- (b) (i) black; 1
- (ii) $X^G X^g$;
(lose this mark if the wrong genotype is given for the female in (iii))
(must show X chromosomes to gain the mark) 1

correct parent gametes
(X^g and Y from male, X^G and X^g from female);
correct offspring genotypes ($X^g X^g$, $X^G X^g$, $X^G Y$, $X^g Y$);
correct link of offspring genotypes with phenotypes;
 $X^g X^g$ black female
 $X^G X^g$ tortoiseshell female
 $X^G Y$ ginger male
 $X^g Y$ black male
(correct gametes, offspring genotypes and link with phenotypes based on incorrect parent genotype = 3 marks) 3

- (c) $X^g Y dd$;
correct male kitten genotypes ($X^g Y Dd$ and $X^g Y dd$);
correct link of kitten genotypes with phenotypes;
(ignore female kittens)
- $X^g Y Dd$ black
 $X^g Y dd$ grey
(correct kitten genotypes and phenotypes based on incorrect parent genotype = 2 marks) 3

Q46.

- (a) sandy stated as heterozygous / suitable allusion to alleles;
suitable cross chosen; (as in table)
N.B. second two points linked, not stand-alone
- explained why could not be codominance;
N.B. Second two points linked, not stand alone

Suitable cross	Reason why not codominance
3 and 4	Offspring should all be sandy

10 and 11	Offspring should all be sandy
7 and 8	Offspring should all be red

BUT if candidate assumes sandy is homozygous, mark accordingly e.g. "look at cross 1 and 2; all their offspring would be sandy;" and not that, if red or white then identified as heterozygote, then full 3 marks are still possible.

3

(b) 11 aabb,

10 = AaBb, (N.B. only possibility, not A-B-)

2 = A_bb or aa B- (or one possible genotype);

if all 3 correct - 2 marks / if 2 correct - 1 mark; one or fewer - 0 marks

2

(c) 1 mark for each element of clear explanation i.e.

- choice of a suitable piece of evidence;

- explaining why Hypothesis 2 could not account for the observed result;

(only cross really possible is 1 and 2) i.e. if sandy was aaB_, individuals 1 and 2 would both have been aaB; so their offspring could only be either white or sandy (as no A alleles present);

2

(d) (Mark line by line, not to 'first error': do not allow for consequential errors)

Individual 18

Other parent

Parental
genotypes

AaBb;

*No mark for this
(AaBb)*

Parental gametes **AB Ab aB ab**

and

Ab ab;

Offspring
genotypes

AABb	Aabb	AaBb	Aabb
AaBb	Aabb	aaBb	aabb

(Punnett not necessary)

Offspring
phenotypes

red

sandy

white

Expected ratio

3

4

1;

4

[11]

Q47.

- (a) (Gene 1) allele A makes enzyme converting J to K / colourless to red;
 Allele a produces no / non-functional enzyme;
 (Gene 2) allele B makes enzyme converting K to L / red to purple;
 Allele b produces no / non-functional enzyme;
 ("Recessive alleles produce no / non-functional enzyme" = 2)
 White flowers result from genotype aa;
 ... regardless if B or b / even if aaB_
 Colourless (substance) / J produces white;
 Red flowers when A_ bb / enzyme 1 only;
 Purple flowers when A_ B_ / enzymes 1 and 2;

6 max

- (b) (i) (1) (red parent) AAbb;
 (2) (white parent) aaBB;

2

- (ii) F₁ are AaBb;
 F₂ ratio of 9 : 3 : 4;
 Purple : red : white;
 Suitable working shown;

4

- (c) (i) aabb, aaBb, and aaBB; (allow aabb & aaB_)

1

- (ii) (Crush each type of white petal to make an extract, and) add some of
 the (red) pigment / K, to petal OR incubate with K;
 (extract becoming) purple is identified as aaBB OR that staying red, after
 K is added, is aabb;

2

[15]

Examiner reports

Q1.

- (a) Approximately 72% of students gained at least one mark for this question. The most common correct responses related to linkage and the random fusion of gametes. A significant number of students mentioned small sample size. Epistasis was infrequently suggested. The most common incorrect response was 'mutation', followed by environmental factors and epigenetics.
- (b) The great majority of students (83%) provided the correct genotype of *ttmm*. Variations such as *mtmt* were also credited, but students should be encouraged to use the standard format. The most common incorrect response was *tm*.
- (c) This question tested a new topic on the specification and proved challenging for many students. Despite the stem of the question and the figure providing evidence of linkage, this was not mentioned by many students in their explanations of the results of the genetic cross. This resulted in 45% of students scoring zero. Students who did refer to linked genes did not always mention crossing over or the low number of phenotypes produced because of this. Consequently, only 31% of students obtained at least two of the three marks available. The best answers explained that if the linked genes were inherited together, the offspring would be in a 3:1 ratio, but that crossing over produced small number of *Tm* and *tM* gametes that resulted in the low numbers of recombinant tall and mottled, and dwarf and normal plants. Many students who scored zero suggested that the ratio should be 9:3:3:1 and explained why the plants with the dominant phenotypes were most frequent. A few students suggested that mutations had caused the phenotypes which occurred in low numbers. Some students suggested that crossing over occurs in mitosis.
- (d) Almost 50% of students obtained both marks for this question. Students gaining a single mark (30%) usually correctly completed the phenotypes of the offspring in **Table 2**. Incorrect responses for the ratio of offspring in **Table 2** varied considerably, however a ratio of 1:1:1:1 was seen relatively frequently.

Q2.

- (a) Despite less than 13% of students obtaining more than two marks, this question proved to be an excellent discriminator. The most frequently awarded marking point was that the frequency of the *LP* allele would increase in the offspring generation. Many students who gained this mark often gained credit for stating that individuals with the advantageous allele would survive and reproduce. However, poor terminology, particularly referring to 'gene' rather than 'allele', prevented a significant number of students from gaining this mark. There were also many references to 'alleles surviving and reproducing'. Although many students realised that drinking milk gave a nutritional advantage, a named nutrient was not always included to gain a mark. Similarly, mutation was mentioned by some students but this was often not credited as many of these responses suggested that drinking milk had caused the mutation. Directional selection was only included in a minority of responses. A significant minority of students thought that *LP* occurred in cattle and suggested that cattle were selectively bred to continue lactase production. Some students also suggested that the allele for *LP* was obtained from cow's milk.
- (b) Approximately 50% of students gained a mark on this question, invariably for referring to *LP* as a dominant allele. Half of these students obtained the second mark by explaining that dominant alleles were expressed (in the phenotype). Many students who scored zero simply suggested that selection for the *LP* allele caused

its rapid rise, indicating they had not carefully read the question. Perhaps of greater concern was the number of students who suggested that the recessive allele of the *LP* gene had reverted to its dominant form either via mutation, methylation or acetylation.

- (c) As expected, this proved difficult for many students with only 18% obtaining at least one mark and 5% gaining both marks. Many students had no idea how a mutation in DNA could cause lactase persistence. There was often confusion between lactase and lactose. Misconceptions included: mutations in proteins, mutations causing new genes to code for lactase and epigenetic changes/methylation/acetylation initiating lactase production. Some responses described deletion mutations in a different gene on the same chromosome, resulting in a frameshift which affected the lactase gene. Students who had some idea did write about mutations in a gene that codes for the protein that switches off lactase. Others mentioned transcription factors, but the mutations were often in the transcription factor rather than in the gene coding for it.

Q3.

- (a) The definition of phenotype was generally well known with over 90% of students obtaining at least one mark. Students scoring zero usually provided a GCSE standard answer in terms of the phenotype representing an observable feature. Responses awarded a single mark usually described the genetic influence on the phenotype. Better answers also included the effect of the environment, to gain both marks.
- (b) Almost 60% of students correctly named the type of gene interaction shown in the diagram as epistasis. Common incorrect responses included dihybrid and codominance.
- (c) Almost 85% of students correctly identified the genotypes provided as phenotypically white and yellow respectively.
- (d) Over 55% of students obtained all three marks in this question which proved to be a very effective discriminator. However, it was surprising to note that over 25% of students scored zero marks on what was considered to be a relatively straightforward genetic cross. It was difficult to give any credit to students who started their answer by providing incorrect genotypes of the offspring. A number of students were unable to determine the correct phenotypes despite having provided the correct genotypes. A few gave the correct genotypes and phenotypes, writing white, white, yellow, green but then gave a 1:1:1:1 ratio. The ratio was also sometimes given as 9:3:3:1 even though the genotypes and phenotypes were correct.
- (e) Approximately one in six students obtained both marks for this question. This low success rate was not due to a lack of mathematical ability, but due to misinterpreting the information provided. Most students mistakenly thought that 36% represented the homozygous recessive genotype and obtained the incorrect answer of 48%. These students often obtained one mark as they showed that $2pq$ represented the frequency of the heterozygous genotype. Students who realised that that 64% represented the homozygous recessive genotype generally obtained both marks, although occasionally an answer of 0.32 was given which was awarded one mark.

Q7.

- (a) This was well answered with almost 60% of students obtaining both marks and less than 5% scoring zero. Students who only obtained one mark often referred to the

influence of the genotype/genes/alleles on the phenotype but omitted any reference to the effect of the environment.

- (b) (i) Four out of every five students obtained at least one mark on this question usually for correctly identifying individuals 2 and 4 or 10 and 12. However, many of these students incorrectly stated that both parents ie 1 and 2 or 10 and 11 would be heterozygous, having failed to note that the condition is sex-linked. However, almost 40% of students did realise this and gained both marks by indicating that only the female parent would be heterozygous.
- (ii) Almost 80% of students correctly gave the genotype of individual B as $X^B X^b$.
- (c) (i) 40% of students correctly calculated the probability of producing a colour blind, non-tongue-rolling son as 0.0625 (6.25% was accepted). Approximately 5% of students gained one mark for the answer of 0.03125 (or 3.1%), achieved by multiplying the correct answer by 0.5. A common incorrect answer was 0.25 (or 25%) due to students only working out the probability of one outcome.
- (ii) This caused few problems with almost 70% of students obtaining both marks and 90% at least one mark. Students gaining a single mark often provided an incorrect answer but provided evidence of attempting to calculate $2pq$. A small minority of students were awarded a single mark for providing their answer as a frequency, ie 0.48.

Q8.

- (a) This question discriminated more than expected, with 34% obtaining two marks and 50% one mark. Most students wrote about ATP being used to make/break the binding with actin. A few failed to make any reference to actin and failed to score. Those who obtained the second mark went on to refer to ATP providing the energy to move the myosin head.
- (b) (i) 63% of the students could calculate the correct percentage fall.
- (ii) 63% of students obtained both marks and 30% obtained zero. Those who failed to score often wrote an account of what phosphocreatine is used for. 'These results' they were asked to explain included the mice being unable to produce creatine.
- (c) 74% obtained one mark for the observation that the heterozygous mice have one dominant allele (of the gene for creatine production). Only 11% went on to say that this one allele allowed the mice to produce enough, or the normal amount of, creatine. Some said that the dominant allele masks the recessive allele, which is true but doesn't explain why the heterozygous mice can produce a similar force to normal mice.

Q9.

- (a) Over ninety percent of students provided a suitable definition of codominance although the quality of expression varied considerably.
- (b) Again this was a well answered question, with over eighty percent of students obtaining the mark. Correct responses usually referred to males possessing only one allele (of the gene) for fur colour or that males could not be heterozygous for this condition. Explanations that focused on the Y chromosome often failed to gain the mark. A few answers only explained that males were XY and females XX without any reference to the alleles (of the gene) for fur colour.

- (c) Almost ninety percent of students gained at least two out of the three marks for this question by correctly carrying out the genetic cross and providing the correct genotypes and phenotypes of the offspring. However, just over a third of these students included the sex of the cats when determining the ratio of the phenotypes to obtain maximum marks. Some students did not provide ratios but gave percentages or fractions. A minority of students did not seem to understand the terms genotype and phenotype. Very few students scored zero.
- (d) (i) Surprisingly, fewer than thirty percent of students correctly calculated the frequency of the recessive allele as 0.9. A number of students provided an answer of 90%, without always showing the working, to gain at least one mark. A common error was to use 19% as the frequency of the dominant allele, p and then subtract this from 100 to give 0.81 as the frequency of the recessive allele, q . Another incorrect calculation used 0.19 as q^2 and then q as 0.44, subtracting this value from 1 to give an answer of 0.56.
- (ii) This was not well answered with less than one in four students obtaining the mark. Many students referred to the homozygous dominant genotype increasing in frequency without any mention of the homozygous recessive genotype. Other incorrect responses suggested that both homozygous genotypes would increase, discussed only allele frequencies, or confused allele and genotype. Some answers compared frequency of homozygous with heterozygous genotypes or referred to the allele for polydactyly becoming 'more dominant' or 'recessive'.

Q10.

- (a) Over half of students managed to communicate the idea that the same restriction enzymes cut at the same place / recognition sequence on DNA. Only just over 10 percent considered the context of the question and went on to say that this would give fragments containing the same gene (R).
- (b) About half of students obtained both marks in this part and over 40 percent obtained 1. Most managed to convey the idea that there was binding between complementary base sequences on a primer and DNA from an allele. The better answers conveyed the idea of a specific base sequence for each primer.
- (c) Some very good and clear answers were seen to this part and a third of students obtained 3 marks. These students identified the genotypes of L, M and N and explained how they identified them on the basis of the sizes of primer attached to each and how far the bands moved. Quite a large number went on at length about the offspring as represented by M but failed to identify L and N. A few thought that there were two types of offspring because there were two bands for M; they clearly did not understand the simple genetics of the cross.
- (d) This proved far more challenging than expected. The examiners were looking for the idea that chromosomes would be visible (as separate structures) and thus the scientists could see to which chromosome the probe was attached. Fewer than 10 percent got both marks.
- (e) (i) There were quite a large number of attempts at generic 'How Science Works' answers to this part. 50 percent of students obtained 1 mark, usually for suggesting that the control was to see the effect of the insecticide or that it was for comparison with resistant flies. However, only around 10 percent gave both of these.
- (ii) Most students focused on one aspect of the results and scored 2 marks. They

frequently spotted that if the enzyme was the only factor in resistance, then the results for the controls and the resistant flies with the inhibitor would be the same. Others focused on evidence for the statement by comparing the resistant flies with resistant flies with the inhibitor added.

Q11.

- (a) Almost two thirds of students gained this mark, most often for stating that females only needed to have one copy of the recessive allele for it to be expressed. Other students referred to males requiring two recessive alleles for the recessive phenotype to be expressed. Unfortunately, some students did not use the correct terminology, with gene or chromosome being used instead of allele. Sometimes, the information that in birds the female is XY and the male XX was forgotten, so the answer was the wrong way round.
- (b)
 - (i) The majority of students did use the correct evidence from the figure, ie 1, (2) and 5, and gained one mark. However, only the better answers explained that parent 1 was heterozygous or a carrier; many referred incorrectly to both parents 1 and 2 or to one of the parents being heterozygous without specifying which. Individual 3 was also frequently included as a child of 1 and 2.
 - (ii) Two thirds of students gained both marks for this question. Students who scored one mark usually did so for correctly identifying the possible genotypes of chicken 7. Common errors were to provide autosomal genotypes or to show an allele on the Y chromosome of chicken 5.
 - (iii) Just over 50% of students gave the correct genotypes of the parents. As with b(ii), a number of students provided autosomal genotypes.
- (c) Two thirds of students obtained all three marks. Few students were awarded two marks but a significant percentage scored one, often for identifying the heterozygous genotype as $2pq$. One common error was that p / q , rather than p^2 / q^2 , was often identified as 0.04.

Q12.

- (a) The phenotype as the expression of the genotype was well known, with the vast majority of students gaining the first marking point. Less than a quarter of students mentioned the effect of the environment on the phenotype. Answers that failed to gain a mark just described the phenotype as the visible characteristics or appearance of an organism.
- (b)
 - (i) Over a third of students scored zero on this question. Most of these students suggested that the phenotypes of 1, 2, and 4 / 5 proved that the allele for Tay-Sachs disease is recessive. There was also a common misconception that individual 3 was the son of 1 and 2. Approximately half the students who correctly referred to individuals 3, 4 and 9 / 11, provided a suitable explanation to gain a second mark. Unfortunately, the use of poor terminology prevented more students from gaining both marks; for example, 'do not carry the gene'.
 - (ii) This proved to be very problematic with less than twenty percent of students scoring above zero. Many students had no idea how to prove that the allele for Tay-Sachs was not on the X-chromosome, often identifying the wrong individuals or suggesting that only males should be affected. Some students gained the first mark for correctly selecting individuals 11 and 3, but only a minority of students were able to explain how this proved that the allele was not on the X-chromosome. The allele was often thought to be on the Y-

chromosome. The fathers were also thought to pass their X-chromosome to their sons. Some answers were very general and failed to mention any numbered individuals.

- (c) (i) This proved to be quite an effective discriminator despite fifty percent of students obtaining all three marks. Students obtaining a single mark often did so by attempting to calculate $2pq$. Although many students gained credit for showing that $q^2 = 0.001$, a significant number incorrectly indicated that $q = 0.001$. It was disappointing to find incorrect answers which showed no 'working' as some of the calculation might have been along the right lines. The required use of decimals certainly proved troublesome for some students. A small minority of students simply provided the Hardy-Weinberg equation but made no attempt to use it.
- (ii) It was surprising to find that almost fifty percent of students did not gain this mark. Most correct responses noted the information provided in the stem of part (b), ie 'sufferers of this disease often die during childhood'. Incorrect responses related to more general reasons why the Hardy-Weinberg equation might not apply, such as emigration, immigration, lack of random mating etc. Random fertilisation and mutation were often seen as incorrect responses. Natural selection, selection against individuals with Tay-Sachs, stabilising selection and directional selection were also suggested.

Q13.

- (a) The question required the calculation of a ratio and not the statement of the ratio as 142:50. A few did not follow the wording of the question and incorrectly gave the ratio as 1:2.84.
- (b) Most students were able to score both marks with basically the same idea – not being able to tell one form from another – suggested in each case. Some assessors gave credit to references to amputation or deformities, which were not included in the Marking Guidelines and were inappropriate here.
- (c) Many students were prepared to accept the percentage given without considering how it might have been derived. As a consequence, few were able to achieve both marks. Where credit was given, it usually related to a small sample size or the location not being representative of the UK as a whole. It was rare to see a student offer both of these ideas.

Q14.

- (a) (i) A large number of students had difficulty translating the relevant Figure into a description of results. There was a tendency to quote figures without identifying what was shown such as the lack of an overall pattern, or that there was little difference between certain islands.
- (ii) Only the higher scoring students were able to gain more than one mark. The nature of the question was "Suggest" but few could use prior knowledge or understanding in the context of island populations beyond the idea of less, or no, immigration or emigration.
- (b) This question, testing the ability to interpret given evidence, proved to be challenging for students. Few achieved better than a single mark and this was often the result of a lenient interpretation of the Marking Guidelines by the assessor.
- (c) The majority of students could suggest the evidence that would support the

conclusion, which was a complete contrast to the limited ability to interpret evidence shown in the previous question.

Q15.

- (a) Two out of three students gained at least one of the two marks, often for linking the large number of eggs to a large sample size. Approximately twenty five percent of students obtained the second mark, usually by suggesting why the small size or distinct markings made the fruit fly a useful organism for studying genetic crosses. A third of students scored zero as they often provided two features of the fruit fly but no explanations of how these features were useful.
- (b)
 - (i) Almost seventy percent of students obtained full marks. Students scoring zero often completed an autosomal cross with no reference to the sex chromosomes, or showed an allele for eye colour on the Y chromosome. Students gaining two marks often showed the red-eyed males with a white-eyed genotype.
 - (ii) The most common correct responses related to random fertilisation or a small sample size. Slightly more than half the students did not gain the mark, with many using the terms random, or chance but not in the correct context.
- (c) Most students gained at least one mark for explaining that males have one allele that determines their phenotype. Better answers then provided a variety of suitable explanations as to why females were less likely to express a phenotype controlled by a recessive allele on the X chromosome. Weaker answers often referred to dominant or recessive genes / chromosomes, or used incorrect terminology, e.g. homologous rather than homozygous.

Q16.

- (a) The vast majority of students gained the mark for explaining what is meant by a dominant allele. Unfortunately some students suggested that it is 'expressed in a gene'.
- (b) Most students were able to provide all the correct genotypes which would result in a brown-shelled snail.
- (c) This also proved to be a high-scoring question with many students gaining all three marks. Students gaining two marks often failed to link correctly the genotypes of the offspring to their phenotypes. A small minority of students misread the question and completed a genetic cross between a yellow snail and a pink snail. If this was done correctly, one mark was awarded. Very few students scored zero.
- (d) This proved far more demanding. The most common error was to have $q^2 = 0.51$ rather 0.49 as students assumed that 51% represented the homozygous recessive snails, rather than the combination of the homozygous dominant and heterozygous snails. Another incorrect method involved taking 0.51 as the frequency of the dominant allele (i.e. p) and 0.49 as the frequency of the recessive allele (q). These errors often resulted in only a single mark being awarded.

Q17.

- (a) The answer to part (i) was very clearly and concisely expressed by the more able students, especially if they only considered the dominant/recessive nature of the alleles. Those who also tried to include the sex-linked aspect were more likely to produce confused responses. Many students gained the first marking point. Without

indicating the number of the individuals referred to, it was difficult to show use of evidence from pedigree. Students who tried to explain the converse, of why hairiness was not dominant, were not able to do so unambiguously. A few students stated that it must be recessive as not many of the cattle were hairless.

The mark for part (ii) was most frequently gained by stating that only males are hairless or no females are hairless.

- (b) The best students gained all four marks. Many gained three when they fully understood and had the correct genotypes and gametes but, unfortunately, omitted to include the sex of the offspring. A significant number of students managed to gain three marks by carrying out an autosomal heterozygous cross. Quite a few gave up after trying to complete the parental genotypes. In some cases, the X and Y chromosomes were included in the parental genotypes, then missed out in the gametes, only to return again in the offspring genotypes. The fourth marking point was sometimes gained in isolation. The Y chromosome was sometimes shown carrying an allele. Many students made errors because they were unable to read their own writing, mistaking a tiny superscript H for h.

Q18.

- (a) Many of the explanations given in part (i) were very superficial, for example “fewer white kittens so it must be recessive”. References to genotypes were required, although the term carrier was accepted as synonymous with heterozygote. In part (ii), it was evident that some candidates clearly did not understand the term ratio. An understanding of ratios is a mathematical requirement stated in section 3.9 of the specification.
- (b) Most candidates gained the mark for part (i) but part (ii) proved more challenging. A common mistake, perhaps because the sex of the parent cats had been stated, was to assume that the inheritance of this fur colour was sex-linked. Another common error was to miss out the Bb genotype for the black offspring. There were many good answers to part (iii) although few candidates commented that gametes are not always produced in equal numbers, as the Mendelian ratio assumes, or that a small sample was involved. In part (iv), many candidates assumed that they were only expected to suggest a single cross that would produce all chocolate cats, rather than produce a self-sustaining population. Nevertheless, a pleasing number thought through the problem carefully with many mentioning all the marking points in their answers.

Q19.

- (a)
 - (i) The majority of candidates gained the mark for explaining what is meant by a recessive allele. Unfortunately, some candidates simply stated that it is ‘not expressed in the phenotype’.
 - (ii) Again, this was well answered with most candidates expressing themselves clearly with appropriate scientific terminology. Incorrect responses suggested that codominance indicates different genes.
- (b)
 - (i) Surprisingly, only one in five candidates gained any marks for this question. Most candidates suggested that parents 1 and 2 having produced a rhesus positive child was evidence that the allele for Rhesus positive is dominant. It should also be noted that many candidates suggested that 4 was the child of 1 and 2, indicating a lack of understanding of family trees.
 - (ii) Again, candidates struggled to gain any marks. A significant number simply

stated that as males and females have the condition it can't be on the X chromosome. A similar number of candidates suggested that the gene was carried on the Y chromosome. Candidates who did gain credit often referred to 3 being Rhesus positive as evidence that the gene is not on the X chromosome. Fewer candidates cited 9 being Rhesus negative as evidence. Very few candidates were able to provide a suitable explanation to gain both marks.

- (c) Almost half the candidates gained all three marks. The most common error was to assume that $q=0.16/16\%$ rather than q^2 . However, most of these candidates still gained a mark for indicating that $2pq$ represented heterozygotes. A significant number of candidates gained two marks for the answer 0.48.

Q21.

- (a) Many candidates produced stock answers to the question 'describe how meiosis takes place'. By not concentrating on the key features, candidates are likely to produce responses that address those features in insufficient detail. Good candidates, however, were more selective and produced focused responses which described how crossing over between the non-sister chromatids of bivalents produced new combinations of alleles and how random segregation of homologous chromosomes reduced the chromosome number and produced different combinations of chromosomes. They usually also went on to describe the role of meiosis II in splitting the chromatids. A common omission from otherwise good answers was to fail to explain how the processes of crossing over and random segregation introduce variation.
- (b)
 - (i) Good candidates made use of all the information supplied and were able to deduce that to produce black offspring the white parents must be $aabb$ not $AAbb$ and the agouti mice must be Aa not AA ; to produce white offspring the agouti parents must be Bb , not BB . The good candidates then laid out the cross correctly and were able to show the correct ratio of genotypes. Some failed to gain a mark by not clearly showing which genotype produced which phenotype. Candidates who did not make use of all of the information, frequently began from wrong parental genotypes, and often ended with the wrong ratio. Oddly, they did not then attempt to correct this.
 - (ii) Most candidates were able to calculate χ^2 correctly, although many made a basic arithmetic error in calculating $(O - E)$ for the white mice. These candidates correctly calculated E as 60, but then in subtracting 51 from 60 produced the answer 11.
 - (iii) Most candidates correctly stated that there would be two degrees of freedom. They were then able to correctly relate **their** calculated value of χ^2 to the critical value at a probability level of $p = 0.05$ and, from this, to correctly infer whether or not the results were, or were not, significantly different from the expected ratio.

Q22.

- (a) The majority of candidates were able to complete the table correctly to show the missing levels of classification of the guinea pig.
- (b) In explaining sympatric speciation, many candidates demonstrated that they were well versed in the various forms of isolating mechanism, although some confused sympatric and allopatric speciation, and suggested various inappropriate geographical forms of isolation. Many also appreciated that changes in allele

frequency or divergence of gene pools might occur between separate populations. Many forgot to mention *mutation* as the cause of changes in alleles.

- (c) Careless handwriting by many candidates made it difficult for examiners to distinguish between letters used as symbols for the alleles. Although a fair proportion got this section completely correct, errors included monohybrid crosses and sex-linkage.
- (d) The vast majority understood the meaning of the term *null hypothesis* and were able to state a suitable one.

Q23.

Unit 6

Most candidates were able to cope with the genetics of colour blindness but the setting out of the genetic diagram often left much to be desired in terms of clarity of presentation. Those opting for the Punnett square approach typically omitted the parental genotypes and often failed to relate genotype and phenotype for the colour-blind male offspring. Those choosing to use line diagrams often omitted the gametes. A small minority did not understand proportions and could not distinguish between % and 1:4, or between 1:3 and 3:1. Some multiplied their correctly derived answer by 'A to allow (again) for the child being male.

Unit 7

It was pleasing to see that many candidates could produce a genetic diagram that enabled them to score of at least 2 marks, with better candidates scoring maximum credit. The parental genotypes and the probability value were usually correct, although several omitted the parental genotypes when using a Punnett square. Common errors included omission of the gametes where direct lines between parental and offspring genotypes were used, or a failure to identify the male with red-green colour blindness. Giving a ratio of 1:4 or 3:1 and stating the male parent's genotype as X^cY were less frequent but still quite common errors. Several candidates multiplied their correct probability of % by 'A because they failed to realise they had already taken account of the child being male.

Q24.

Unit 8

- (a) Most candidates were able to explain that an increase in water temperature would influence a relevant feature such as oxygen solubility or respiration. Answers based on the effect of temperature on the rate of enzyme activity or on metabolism were, however, a little too general, failing to relate to the specific investigation described in the question. There were a few references to ensuring "a fair test", an entirely inappropriate response at this level.
- (b) The responses to this section formed a sharp contrast to the high marks frequently awarded for statistical analysis in coursework. Answers to part (i) were often centre-dependent, some candidates being able to produce a sound null hypothesis; others clearly had little idea. These candidates frequently lacked understanding of the purpose of the investigation or of the concept of a null hypothesis. The weakest responses usually involved equating the expression with an inappropriate statistical formula. In part (ii), many candidates were aware that statistical tests are related to chance, but fewer were able to explain that such tests give a measure of the probability that chance might account for the results obtained.

- (c) Most candidates correctly identified A as the more likely explanation and were able to justify their choice.
- (d) Better candidates were able to produce in a logical account in which they successfully linked a lower oxygen concentration to anaerobic respiration and the production of lactic acid. Others revealed a disturbing lack of understanding of respiratory biochemistry, suggesting that the evolution of carbon dioxide was entirely independent of the consumption of oxygen. They inevitably based their answers on an argument that, despite reduced oxygen, fish must continue to respire aerobically, so there would be an increase in carbon dioxide. There were occasional references to supposed chemical effects of zinc.
- (e) The best candidates used common sense in part (i) and, realising that the only elements that were concentrated were copper and cadmium, calculated appropriate ratios for these ions. Credit was also given to those who supported their conclusions by calculating the inverse. A significant number, however, merely subtracted the relevant values from each other, an approach which inevitably led to an incorrect answer. The examiners were instructed to be generous in marking the calculations and undertook much work in interpreting confusing presentation. Centres would do well to advise candidates that it is their responsibility to present material sufficiently clearly that the logic of the response can be followed. In part (ii), most recognised that lead ions would be egested or excreted, although there was some incorrect usage of these terms. Most candidates were aware, in part (iii), that woodlice would concentrate copper. The principle of bioaccumulation was often correctly described but not always related to eating a large number of leaves. Weaker candidates frequently referred to additional sources of copper ions or to the intriguing possibility of copper ions multiplying within the body of the woodlouse.
- (f) Mutation figured widely in the responses to part (i), although there were occasional incorrect references to natural selection or to the presence of arsenic causing the allele to first arise. Although there were a number of rather vague references to growth and formation of new cells, the majority of candidates were able to identify two specific effects of phosphates in part (ii). Answers to part (iii) were frequently marred by a failure to answer the question and explain why arsenic-tolerant plants were unable to compete in the conditions described. Candidates referred to both arsenic-tolerant and non-tolerant plants as “they” and it was often far from clear as to which they were referring. However, it was encouraging to note that, although this question was targeted specifically at Grade A candidates, many others were able to suggest that arsenic-tolerant plants would not grow as well because they were unable to take up sufficient phosphates.

Unit 9

- (a) Candidates offered a range of explanations, suggesting that the air mixed the water, that it affected the zinc, and that it was needed to make the test fair. Few candidates earned a mark; those that did suggested that oxygen would no longer be a limiting factor. Links were rarely made with the effect it would have on the saturation of the haemoglobin.
- (b)
 - (i) A large number of individuals know from their coursework that the term ‘null hypothesis’ implies ‘no difference’, but they did not always recognise where this lack of difference might lie. Weaker candidates made comments about chance. The commonest error was to devise a hypothesis relating to gas exchange and respiration.
 - (ii) Many commented on the need to look for effects that are due to chance. Some quoted significance levels, but failed to mention probability. Many referred to

establishing levels of accuracy, and a few made statements about the null hypothesis. It was disappointing to note that large numbers of candidates are able to suggest null hypotheses in their coursework but are unable to apply these statistical skills to material presented in an unfamiliar context.

- (c) Most candidates recognised the answer as A, and were able to use the graph to explain their choice. Those that could not were vague in their answers.
- (d) Unless candidates recognised that there was less oxygen available to the cells they were inclined to answer irrelevantly. The best recognised the anaerobic respiration that would ensue, and therefore lactic acid would be produced. Some wrote of haemoglobin as a buffer, but failed to recognise that it would be the extra hydrogen ions which affected the pH not those absorbed through the buffer. Weaker candidates were confused over the numbering of the pH scale. They thought that zinc affected the pH of the water, or that zinc caused haemoglobin to pick up fewer hydrogen ions from the water.
- (e)
 - (i) The calculations were absent in some cases, and very varied where present. Simple ratios were the best idea, but some even calculated standard deviations. Subtractions were also fine. Many candidates had no idea what to calculate. The commonest response was to find the mean concentration of cadmium and copper in shrews, without any reference to the levels in the source of food. Many gave calculations without saying what they were, leaving the examiner to guess. The weakest candidates mis-read the data as numbers of shrews or numbers of ions. Despite poor performances on the supporting mathematics, many candidates could comment on the relative concentrations.
 - (ii) The fate of the ions was mixed. There appears to be widespread confusion over egestion and excretion and the fact that ions have to be absorbed before they can be used appears to have escaped some. Weaker candidates were of the opinion that the copper ions could be broken down.
 - (iii) Candidates had little understanding of the ways in which ions accumulate through diet.
- (f)
 - (i) Most candidates correctly identified mutations as the cause of the allele arising, but some offered a choice to the examiner regarding natural selection.
 - (ii) Likewise, most candidates were able to name two functions of phosphates. A few were confused with protein synthesis. Some answered too vaguely with 'membranes', iii). This part of the question presented difficulties to many and only the better candidates directed their responses appropriately. There were many vague references to "fogs" and inappropriate set-piece answers on inheritance.

Q25.

- (a) A majority of candidates were able to score all three marks available for this section. Nearly all candidates derived correct parental genotypes, although a few made a fundamental error in giving what was, effectively, a haploid genotype (AB, rather than AaBb). Some then lost their way because they wrote down four gametes for each parent - two of each genotype. They then produced a 4 x 4 Punnett square. Because previous experience of this had always produced a 9:3:3:1 ratio, they assumed it would here also.
- (b) Again, a majority produced the correct answer of a frequency of 0.6 for the **a** allele. Some candidates, however, correctly identified the combined frequencies of the rose

and single-combed chickens as q^2 , with a frequency of 0.36. They then obtained q by taking the square root of this to obtain 0.6. Perhaps because they have often been required to find the frequency of the heterozygote, they did this next and offered 0.48 as their answer. This is another instance where not reading the question carefully may have cost marks.

Q26.

- (a) (i) Few candidates appreciated that, although the inheritance of ABO blood groups is an example of multiple allele inheritance, just two of the relevant alleles are present in all the diploid cells of an individual; one on the paternal chromosome and one on the maternal chromosome of the homologous pair. Some thought that there would be three, whilst others thought that there would be none, as 'the antigens are only displayed on the red blood cells'.
- (ii) A surprising number of candidates confused antigens with alleles and wrote IA and IB instead of A and B.
- (b) A good number scored full marks on this part of the question. They deduced that the genotypes of the parents must be IAIO and IBIO and, therefore, the probability of producing a child with blood group A would be 0.25. Some candidates did not take account of the fact that only half the children would be male.

Q27.

- (a) Although the definition in (i) produced a variety of responses, ranging from the short and precise to the lengthy and rambling, most were acceptable. Part (ii) was straightforward but two common errors were made, sometimes by good candidates. These were to give respiration or urination as responses. The hypothalamus was correctly identified by most candidates.
- (b) Part (i) was well answered though it appeared some candidates may not have noticed the presence of this question at the bottom of the page. Candidates did not usually find (ii) easy. Many used almost all the space writing about ADH without using the required information from **Figure 1**. Where the role of the aquaporins was understood it was not always made explicit in answers. Osmosis was usually considered and most candidates also appreciated that the aquaporins are bound to the membrane facing the lumen of the collecting duct. However, answers which detailed the whole process were few and far between.
- (c) Unfortunately, some candidates gave the impression of applying a prepared answer relating to the presence of glucose in the urine to this question and, obviously, gained no credit. The mark scheme allowed for all the major obvious symptoms and many of the better candidates obtained the mark. Part (ii) produced good responses from the stronger candidates but very commonly weaker candidates did not appear to know that males had one X and one Y chromosome while females had two X chromosomes. The responses to (iii) were reasonable. The most common shortcoming was a failure to write in terms of probabilities with many candidates stating no more than that carriers of a recessive allele would reproduce while most of those with a dominant allele definitely would not reproduce. A handful of candidates thought that the question was looking for heterozygote advantage and answered in these terms.

Q28.

Many candidates from across the ability range were able to complete the genetic diagram and gain full marks. Some did not realise the dihybrid nature of the cross and some did

not always make clear the relationship between offspring genotypes and phenotypes.

Q29.

- (a) Most correctly pointed out that sex-linkage was indicated in the pedigree diagram by the fact that the condition was seen only in males. Many made the false assumption that being expressed by the minority demonstrated that the allele was recessive; better candidates realised that if the trait was not expressed in the parents but did appear in the offspring then its allele must have been recessive. Some were non-selective in their use of evidence and forfeited the mark. Many insisted on including underlying theoretical points which were not necessary as they did not constitute the *evidence* (i.e. observations) required by the question.
- (b) Most candidates identified the possible genotypes of all four individuals correctly, including the two possibilities for person 7. Unfortunately, many were rather careless with the final genotype, assuming it to be the same as for number 7 (i.e. disregarding the extra information given in the diagram).
- (c) Many calculated the probability correctly as 0.25, often using the extra space available on the page to draw genetic diagrams to verify their ideas. Some spoiled their answers by giving extra, incorrect alternatives – thus, ‘25% or 1:4’ scored zero. Others did not appreciate the difference between ‘1:3’ (correct) and ‘3:1’ (incorrect).

Q30.

Many candidates found this to be a demanding question.

- (a) Most could successfully explain that a clover plant of genotype aaLi would be acyanogenic either in terms of enzyme A not being produced or because the precursor could not have been converted to the intermediate substance linamarin.
- (b) Although many were able to state the four correct gamete genotypes, a high proportion gave them as diploid rather than haploid, or had some containing just one of the two genes and some the other. Some even included X and Y chromosomes in these plant gametes. A significant proportion who failed to give the correct gametes in this section subsequently went on to do so in part (c). Explanations of how meiosis could have produced these types of gametes were usually very weak. Answers tended to be general rather than being applicable to the given situation. There was little mention of the separation of homologous chromosomes and, where independent assortment was included it was frequently negated by a reference to crossing over (ignoring the information given in the stem of the question which stated that the two genes concerned were located on *separate* chromosomes).
- (c) The majority of candidates were able to construct a 4 × 4 Punnett square to show the derivation of offspring genotypes from doubly heterozygous parents. Some, due to careless handwriting, failed to distinguish the symbols ‘L’ and ‘l’ with sufficient clarity. Some failed to indicate which three cells in their table represented genotypes of plants which were acyanogenic and yet able to produce linamarin. Some selected the wrong three (or even four) genotypes. And, while many correctly gave the probability as 3 / 16, some just stated ‘3’.
- (d) Many candidates were unable to state a null hypothesis. Others chose the wrong feature - e.g. stating there was no difference between the number of damaged and undamaged plants (rather than there being no difference between the damage to cyanogenic and to acyanogenic plants). Conclusions drawn from the fact that the probability in the χ^2 test was less than 0.05 were frequently either incomplete or confused. While many appreciated that this indicated a significant difference, not all

rejected the null hypothesis and even fewer related it to the biological situation by stating that being cyanogenic did appear to protect the clover plants from slug damage.

- (e) In this section, candidates' explanations of the data generally got no further than the observation that both types of clover plant tended to thrive at low slug densities while only the cyanogenic survived at high slug densities. Little reference was made to the varying selection pressures operating at the different slug densities and most forgot to mention that cyanogenic clover plants were able to defend themselves against slug damage due to the release of cyanide – hence the selective advantage experienced by these plants if slugs were exercising a significant influence.

Q31.

Whilst it was pleasing to see a few excellent candidates achieving full marks with this question, it was more common to see low-scoring answers. Most candidates still struggle with numerical and statistical treatment of data and its application to biological situations.

- (a) Most candidates gave an appropriate genotype and recognised the approximate 3:1 ratio in the offspring as the evidence for such. A number incorrectly used different letters for the dominant and recessive alleles or assumed the characteristic to be sex-linked.
- (b) Statistical tests are used to test the *probability* of results being due to *chance*. Whilst the concept of chance was well demonstrated, the probability of this being the case was seldom identified.
- (c) Well-prepared candidates were familiar with a definition of *codominant*. Answers were then clear and concise – both alleles would be expressed in the phenotype of a heterozygote. Where this was not the case, weak explanations, such as 'neither is dominant', often failed to make the point.

The clue to the calculations was in the wording of the questions. Part (iii) required use of the Hardy-Weinberg equation. This was not requested in part (ii) and attempting to do so was an inappropriate strategy. All that was required was an addition of all alleles present (1000) against which the *actual* frequency of the particular allele could then be established (0.25 or 25%). Many candidates failed to recognise the significance of calculating the *actual* frequency (250 in 1000).

For most candidates, their remaining credit was restricted to identification of the Hardy-Weinberg equation. Those that were able recognised that their actual frequency would be q , (but would work equally as p) from which q^2 could be determined and applied to determine the expected number with the genotype in the population (500). It was interesting to note that candidates who performed well with the mathematical nature of this question were not always successful with questions testing biological understanding.

Q32.

This proved a challenging question for many candidates. Most correctly discussed sodium ions not entering or leaving the axon (some by diffusion) thus preventing an action potential / impulse. Some knew the neurotransmitter stopped at the synapse. Many went on to discuss the action potential in the postsynaptic neurone / neuro-muscular junction at length. Quite a few failed to gain any marks by writing entirely about postsynaptic transmission. Very few mentioned motor neurones.

Q33.

It was pleasing to see that many candidates got full marks on this question; however some incorrectly used the B and b alleles as subscripts on the X chromosome.

- (a) (i) A significant minority of candidates gave males as XX
- (ii) Often this was correct, even when the male was incorrect in part (i).
- (b) The genotypes of the parents and gametes were often correct but the genotypes of all the offspring were given without indicating which were males. Often, as a result, an incorrect answer of 1 / 16 (sometimes given as a percentage) was the most common wrong answer.

Q34.

This question was a good discriminator, with the full range of marks being awarded and the mean being close to half marks.

- (a) Most candidates answered this correctly.
- (b) There were many correct answers, but also a significant number who gave only one of the two genotypes. Many also gave genotypes of gametes instead of offspring.
- (c) Many candidates correctly identified the pea comb individuals as the ones which would produce blue eggs, but only a minority of candidates gave a complete answer and explained why. Many suggested various crosses, such as a back cross, or other ways to determine if individuals possessed the B allele, not realising that comb shape would provide this information.
- (d) Many candidates realised that crossing over was important here, but only the better candidates made the connection between the closeness and the likelihood of crossing over between the gene loci occurring. Sometimes the answer for (c) was given in this part.
- (e) Nearly all candidates obtained at least one mark, and most of these scored two. Temperature and diet were most common correct answers. The most common answers which could not be given any credit were age, oxygen and pH. Sometimes answers were too vague, such as 'competition' or 'weather'. Stress was also frequently mentioned without the environmental cause of it. Also factors which were unlikely were given, such as UV light, X rays and radiation.

Q35.

Only the very weakest candidates failed to score any marks, although full marks were relatively rare. The principle of dihybrid inheritance appears to be one that lots of candidates understand very well but application of this to complete a cross involving epistasis caused problems for some.

- (a) The majority of candidates gained this mark.
- (b) Surprisingly few could remember epistasis but went on to gain a mark for an acceptable explanation. Many answers were marred by inaccurate or careless explanations where candidates failed to mention gene expression being influenced by the interaction.
- (c) Most candidates managed to give the correct genotypes even after writing down a muddled set of gametes. The ratio was commonly written as 1:1:1:1; suggesting a

genotypic rather than phenotypic ratio.

Q36.

Most candidates could not apply the information given in the stem of the question in (b) and therefore limited the marks gained to two.

- (a) The majority of candidates gained one mark for naming a process but poor expression marred the answers of weaker candidates, in particular the distinction between allele and gene and the concept of different combinations of alleles.
- (b) Candidates did not have the confidence to answer the question set and gave all possible genotypes and phenotypes. The common error was to produce two different gametes from the male, as the vast majority of candidates failed to use the information given under the diagram in the stem of the question.

Q37.

Generally this question was well answered with most candidates obtaining at least five marks. However, part (d)(i) proved difficult for a significant number of candidates.

- (a) This caused few problems with the vast majority of candidates correctly explaining that a *dominant* allele is always expressed in the phenotype or codes for a functional protein.
- (b) Most candidates correctly named the relationship between the two alleles as codominance. A common incorrect response was epistasis.
- (c) The majority of candidates had little difficulty completing the genetic diagram to obtain all four marks. However, a number of candidates failed to gain a mark for the correct ratio of offspring phenotypes. Candidates failing to gain any marks often attempted a monohybrid cross.
- (d)
 - (i) Only better candidates gained both marks. Common incorrect responses referred to mutations or to sperm being XX or XY.
 - (ii) Although many candidates did refer to obtaining milk or meat, not all candidates linked this to the gender of the cattle. It was disappointing to find a significant number of A level biologists referring to 'milk from bulls'.

Q38.

This question was well answered by the vast majority of candidates and was often the highest scoring question in section A. A significant number of candidates obtained maximum marks.

- (a) It was extremely rare to come across a script on which both parts (i) and (ii) were not correct.
- (b) The vast majority of candidates obtained both marks but some weaker candidates did have problems showing the correct genotypes of the gametes or providing the correct ratio of offspring phenotypes.

- (c) Although many candidates understood the genetic control of coat colour in Labradors, several candidates had difficulty explaining the processes involved. Nevertheless, in part (i), many candidates gained one mark for stating that phaeomelanin would not be converted into eumelanin. Most of these candidates linked this to the lack of an enzyme as there is no dominant E allele present. In part (ii), most candidates gained one mark for stating that the presence of the dominant E allele would result in an enzyme converting phaeomelanin into eumelanin. However, most candidates suggested the black coat colour was due to more eumelanin being produced rather than the idea that more of this dark pigment would be deposited in the coat hairs.

Q39.

There were many excellent answers with a significant number of candidates scoring full marks.

Most candidates scored a mark for stating that males are XY and females XX. There was however considerable confusion between often chromosome, gene and allele, with the terms being used inappropriately. Many of the weaker candidates failed to score the second mark, which related to the chances of a male or female inheriting colour-blindness, due to poor expression.

There were many totally correct answers. Common errors included omitting the H and h alleles, giving only the sexlinked alleles or making the H and h alleles sexlinked. There was some confusion about how many alleles to include in a genotype of a gamete, but generally those who wrote correct genotypes in part (i) were able to follow through to obtain the correct gametes in part (ii) and the correct probability in part (iii).

Q40.

- (a) This question produced the full range of marks.
- (b) This was often poorly-answered, as many candidates seemed to misread or misinterpret the question set, and wrote about the causes of variation or the production of different combinations of alleles. Although these candidates referred to mutation in their answers this was usually in the wrong context so could not be given credit. Very few explained how 'multiple' alleles are produced.
- (i) This was often poorly answered. Answers were often vague or incomplete, just stating that it did not matter what alleles was present with no further explanation in their answer.
- (ii) The majority of correct answers included only two genotypes rather than the four possible ones. Modifications were made to the mark scheme to take into account answers from centres who had not been given the erratum. AQA apologises to centres and candidates for this error.
- (iii) Many referred to the enzyme being denatured at higher temperatures, but did not explain the presence of the pigment at the points, with an active enzyme working at lower temperatures. There was some confusion between enzyme and pigment with many references to pigment denaturation. Weaker candidates tried to explain the differences in terms of camouflage.
- (c) Although many candidates were awarded one mark, many of the answers did not explain the production of the phenotypes fully and just stated that A was dominant over a^h without relating this to production of the enzyme. There was also some confusion between enzyme and pigment here with many stating that A coded for the

pigment, not the enzyme.

Q41.

- (i) Most candidates gained the XX/XY mark, although some used inappropriate terminology describing the chromosomes as genes or alleles. A substantial minority failed to describe the difference between the X and the Y in terms of size.
- (ii) Candidates who gained no marks failed to recognise the condition must be recessive as the mother is described as a carrier. Others failed to recognise the significance of the information given in the stem regarding sex linkage and therefore gained only one mark. There were many inappropriate genetic diagrams (suggesting this synoptic area was not remembered very well) commonly representing the haemophilia allele as H and the normal allele without any symbol, but if a key was included credit was given. It was easier, in this case, to gain marks for a well-annotated genetic diagram rather than using prose but some very good descriptions were seen from the most able candidates.

Q42.

This question produced the full range of marks.

- (a) This part was only answered well by the better candidates. Weaker candidates tended to answer in terms of codominance or dihybrid inheritance.
- (b) This was answered well by the majority of candidates.
- (c) This was only answered correctly by the better candidates. In part (iii) suggestions were often vague, such as use of a mouse with a 'known genotype.' A significant number suggested DNA analysis, which was not given credit.

Q43.

- (a) Many gained full marks. The commonest error in completing the genetic diagram was a failure to distinguish between the gametes for each individual, with the result that they appeared to have an identical genotype to the adult. Some candidates aimed to show only the relevant genotypes for this particular outcome, which was accepted, but then were inconsistent or made careless errors, such as giving the X chromosome instead of the Y for the paternal grandfather. In part (iii), most understood the principle, but there was often careless use of the terms gene and allele.
- (b) Most appreciated the size difference between the chromosomes, but only the best candidates took this further and, for example, explained that chromatids could not line up satisfactorily. A very common misconception was that crossing over would not be possible because of the involvement of 'sex genes'.

Q44.

- (a) This proved to be a discriminating question with few candidates giving the correct diploid number of chromosomes. Common incorrect answers included 46, 23, 12 and 3.
- (b) (i) Most candidates correctly referred to crossing over or a chiasma (although many used the plural, when the diagram showed that a single event was present). A common misconception was observed when candidates described chromosome replication happening in meiosis and also that chromatids pair

together.

- (ii) A significantly high proportion of candidates failed to describe the movement of chromosomes in a way that distinguished meiosis from mitosis. Often, it was difficult to determine whether candidates meant chromosomes were separating from single pairs. Poor expression cost marks for some candidates, such as describing the separation of whole bivalents or the movement of pairs of chromatids.
- (c) (i) Most candidates found this difficult. A common error was to label sister chromatids with different alleles.
- (ii) Very few candidates achieved both marks for this question. Interestingly, a significant number of candidates who were able to give correctly the gametes from a dihybrid cross in the answer to question 5(c) could not extend the principle to three alleles in this question. Often, candidates correctly understood that these gametes contained three different alleles, but could not give the correct total for the number of different types of these gametes.

Q45.

This, for many candidates, was the only question in which they achieved full marks. The role of sex-linkage and dihybrid inheritance appear to be topics that a lot of candidates understand very well.

- (a) Many candidates lost their only mark on this question by failing to give an adequate definition for sex-linkage. Some incorrectly referred to X and Y chromosomes as sex gametes. Others described the location for alleles of a single gene as being on both X and Y chromosomes.
- (b) Most candidates achieved full marks. A failure to make a clear connection between genotypes and the corresponding phenotypes was the most common reason for failing to gain a mark.
- (c) A surprisingly high proportion of candidates correctly used the information given in the stem of the question to determine the genotype of the cream-coloured male cat. Then, almost invariably, they went further to quote the correct genotypes and phenotypes of the male kittens produced in the cross.

Q46.

In this question candidates were presented with a pedigree relating to pig coat colour across three generations. Parts (a) and (c) proved, in general, the most difficult. Most candidates gained at least one mark from part (b), and in part (d) it was not unusual for candidates to gain three or even all four of the marks available.

In part (a) many candidates showed ignorance of the term codominance, which is surprising since it featured in the January module also. Moreover, the question asked for evidence from the diagram (i.e. a particular cross), to show that coat colour was not controlled by one gene with two codominant alleles. It was not appropriate, therefore, to explain instead that two genes would be involved, epistatically, as outlined in Hypothesis 1 on the next page. Correct answers used any single cross (except that between individuals 1 and 2) to demonstrate what would have been obtained had codominance been involved. The third mark could be gained by specifying the possible allelic combinations e.g. RR = red, WW = white, RW = sandy. (Note, however, that where candidates chose to use a different arrangement, such as sandy as one of the homozygous phenotypes, due credit could be obtained in the marking scheme.)

As previously indicated, most candidates correctly gave the genotypes of individuals 11, 10 and 2, as aabb, AaBb, and A_bb or aaB_ respectively. Only a minority of candidates seemed slightly confused by the device used to indicate the presence of either of the possible alleles. For example, A_bb could include individuals with the genotypes AAbb or Aabb. The examiners felt that in such cases the candidates' basic misconceptions in the field of genetics were more fundamental than any confusion this device might have introduced. In part (c), the only cross that could reasonably be used in a correct explanation was that between individuals 1 and 2. The fact that each would lack the allele A in its genotype would not allow the production of red offspring, the genotype of which in Hypothesis 2 does require such an allele. The examiners considered that question parts (a) and (c) allowed candidates a very good opportunity to show what they understood, or did not, of genetics. In part (d), candidates gained marks for identifying the genotype of individual 18 (AaBb), showing the parental gametes, then the offspring genotypes, and finally the expected phenotypic ratio of 3 red: 4 sandy : 1 white. The performance of candidates varied considerably, ranging between those who made no attempt, to those gaining all four marks (and occasionally 3 if they had failed to be sufficiently careful in identifying the phenotypes obtained from the genotypes they had worked out correctly).

Q47.

This question involved epistasis and was based on the effect of two different enzymes in a biochemical pathway determining flower colour. Those candidates who were confident about genetics found little difficulty in gaining all 15 available marks in this question. In some cases, however, candidates answered in terms of monohybrid inheritance, even though two separate genes were clearly implicated. Some candidates gave answers in terms of multiple alleles, or even sex linkage, in which it appeared that they had confused the concepts of allele and gene.

In part (a), candidates could receive full credit by using a number of different approaches to explain how the two genes were involved in producing differently coloured flowers. Some candidates concentrated purely on the alleles and their effect on the two enzymes and the pigments produced, whereas others tackled the question only in terms of the flower colour produced by different genotypes. To gain all six marks available it was necessary for candidates to explain aspects of both.

In part (b)(i), the parents were Aabb (red-flowered) and aaBB (white-flowered). Good answers to part (b)(ii) showed the purple-flowered offspring as AaBb, and proceeded to derive the offspring 2 ratio of 9:3:4, purple: red: white, showing appropriate working. Examiners were surprised and pleased to find that it was not impossible for candidates who had achieved no marks in (b)(i), having offered answers that were totally wrong, to recover and gain full credit in (b)(ii).

In part (c)(i), three different genotypes, aaBB, aaBb, and aabb, were required for the single mark. A proportion of candidates failed to score because they omitted one of these. In part (c)(ii), candidates gained credit by suggesting the addition of the red pigment, K, to extracts of each homozygous type of white petal, going on to achieve full marks for explaining that the extract of the aaBB petal would contain enzyme 2 and that this would catalyse the conversion of K to L, turning the extract purple. In contrast, the other white petal from the aabb flowers would remain red after K had been added. The most commonly encountered wrong approach was for the petals somehow to be 'crossed'.

Genetics questions, as a rule at this level, tend to produce extremes of either very low or very high marks, depending on candidates' understanding of the topic. This question, in requiring quite different skills in its three main parts, proved an exception to this, with many intermediate scores being achieved as well. Where candidates understood the concept of dihybrid inheritance and associated enzymes, answers were very good indeed and showed an improvement in the General standard seen at Advanced level in recent

years.