16. Inheritance

16.2 The Roles of Genes in Determining the Phenotype

Paper 4

Question Paper

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

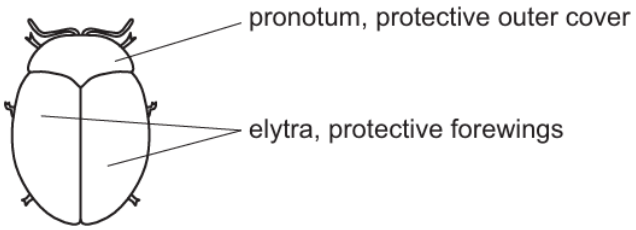


Fig. 3.1

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

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

Table 3.1

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

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

.....

.....

..... [1]

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

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

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

parental genotypes:

gametes:

offspring genotypes:

offspring phenotypes:

ratio of offspring phenotypes:

[4]

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

Use the information in Table 3.1 to:

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

codominant alleles .....

dominance hierarchy .....

[2]

(d) Researchers carried out two crosses.

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

The results are shown in Table 3.2.

Table 3.2

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

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

The results are shown in Table 3.3.

Table 3.3

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

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

cross 1 .....

cross 2 .....

[2]

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

Explain how the evidence in Table 3.3 supports this conclusion.

.....

.....

..... [1]

(iii) Name a statistical test that can be used to determine whether the ratio of observed phenotypes is significantly different from the expected ratio.

..... [1]

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

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

.....

.....

.....

.....

.....

.....

.....

.....

.....

.....

.....

..... [5]

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

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

.....

.....

.....

.....

.....

.....

..... [3]

3 Many different genes are involved in the production of pigments in mammals. One example is the *TYR* gene.

(a) In humans the *TYR* gene is located on chromosome 11.

Fig. 6.1 shows the homologous pair for chromosome 11.

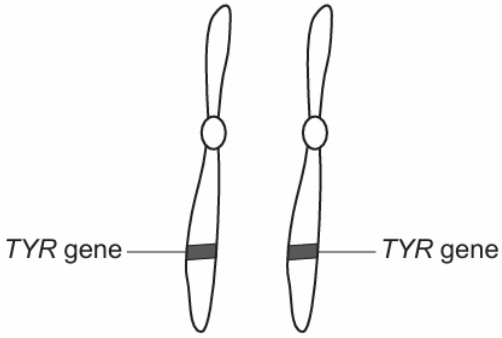


Fig. 6.1

(ii) Albinism in humans can be caused by recessive mutations of the *TYR* gene.

Explain why a person who is homozygous recessive for the *TYR* gene shows albinism.

.....

.....

.....

.....

.....

.....

..... [3]

(b) Fur colour in rabbits involves a number of different genes. Some of these genes interact.

Gene 1 has two alleles, **B** and **b**.

- The dominant allele, **B**, results in black fur.
- The recessive allele, **b**, results in brown fur.

Gene 2 has two alleles, **F** and **f**.

- The dominant allele, **F**, codes for a protein that allows the expression of gene 1.
- The recessive allele, **f**, codes for a protein that does not allow the expression of gene 1, resulting in white fur.

The two genes are on different pairs of autosomes.

Complete the genetic diagram for a cross between two black rabbits that are heterozygous for both genes and show the ratio of possible offspring phenotypes.

parental phenotypes black × black

parental genotypes BbFf × BbFf

Punnett square

ratio of offspring phenotypes:

..... [4]

4 Genetic crosses can be used to investigate patterns of inheritance.

(a) A mutation in a gene involved in fruit colour in tomato plants, *Solanum lycopersicum*, results in the production of yellow fruits instead of red fruits.

A genetic cross was carried out between a pure breeding plant with a red fruit and a pure breeding plant with a yellow fruit to produce the F1 generation. All offspring plants produced red fruits.

The F1 plants were then crossed with each other and the seeds produced were planted to obtain the F2 generation.

Construct a genetic diagram to show the cross of the F1 generation that produced this F2 generation.

Use the symbols **R** and **r** for the alleles.

[3]

(b) A theoretical dihybrid cross involves two genes located on different autosomes. Each gene has two alleles, one dominant and one recessive.

A parent, homozygous dominant for both genes, is crossed with a parent that is homozygous recessive for both genes. This produces F1 individuals that are then crossed to produce the F2 generation.

State the phenotypic ratio of this dihybrid F2 generation **and** explain why some of these offspring phenotypes are different from the original parental phenotypes.

ratio .....

explanation .....

.....

.....

.....

.....

..... [3]

5 Populations of the moth *Biston betularia* live in Europe and in North America. The most common phenotype on both continents is a pale wing colour with light-grey shading (the typical form).

A moth phenotype with dark wing colour (the melanic form) also occurs on both continents.

Fig. 10.1 shows the typical form of the moth.

Fig. 10.2 shows the melanic form of the moth.



Fig. 10.1



Fig. 10.2

(a) Two melanic European moths were crossed together. The wing colours of the offspring were 15 typical and 41 melanic.

Construct a genetic diagram to explain these results. You may use the symbols **A** and **a** to represent the alleles.

[3]

(b) In a similar experiment, two melanic North American moths were crossed together. The colours of the offspring were 10 typical and 31 melanic.

What can be concluded about the allele that causes the melanic form in the moth populations in **both** continents?

…… ..... [1]

(c) Researchers did not know if the allele causing the melanic form in European moths occurred at the same locus as the allele causing the melanic form in North American moths. To find out, they carried out the following crosses:

- Cross 1: European moths that were heterozygous at the European melanic locus only were crossed with North American moths that were heterozygous at the North American melanic locus only.
- Cross 2: The melanic and the typical offspring of cross 1 were mated together.

(i) Explain why cross 2 is a test cross.

….....

….....

..... [1]

(ii) Complete Table 10.1 to show the predicted results if:

- the European and North American melanic alleles are on **the same locus (A/a)**
- the European and North American melanic alleles are on two **different loci (A/a** and **B/b)**.

Table 10.1

	same locus (A/a)	**different loci (A/a and B/b)**
genotypes of **melanic** moths from cross 1		
proportion of test crosses (cross 2) giving 100% melanic offspring		

[3]

6 Domestic rabbits vary in the length and colour of their fur.

Fig. 6.1 shows a domestic rabbit with short fur and a fur colour pattern called Himalayan.



Fig. 6.1

The two genes that determine the length and colour of the fur of this rabbit occur at the **A / a** locus and the **B / b^h / b** locus. These two gene loci are on separate chromosomes.

- The allele **A** results in short fur.
- The allele **a** results in long fur.
- **A** is dominant to **a**.

- The allele **B** results in black fur all over the body.
- The allele **b^h** results in black fur on the nose, ears, paws and tail of the rabbit, and white fur on the rest of the body (Himalayan pattern).
- The allele **b** results in white fur all over the body (albino).
- **B** is dominant to **b^h** and **b^h** is dominant to **b**.

(a) (i) List the **four** possible genotypes of the rabbit shown in Fig. 6.1.

.....

.....

..... [2]

(ii) One phenotype of rabbit always breeds true. This means that when it is mated to a rabbit that looks the same as itself, all the offspring look the same as the parents.

Describe the **phenotype** of the rabbit that breeds true.

..... [1]

(b) A rabbit with long, black fur all over the body that was homozygous at both loci was crossed with a rabbit with short, white fur that was homozygous at both loci. The F1 offspring had short, black fur. These F1 rabbits were mated together to become the parents of the F2 generation.

Draw a genetic diagram to predict the F2 offspring genotypes and the ratio of F2 offspring phenotypes.

F1 genotypes:

gametes:

F2 offspring genotypes:

ratio of F2 offspring phenotypes:

[5]

(c) A rabbit breeder performed multiple crosses of the type described in **(b)**. This gave enough data to test the prediction that the genes for fur length and fur colour show independent assortment.

(i) Explain why the two genes assort independently.

.....

.....

.....

.....

.....

.....

.....

..... [3]

(ii) The rabbit breeder placed the results in a table and started to calculate the χ^2 value.

Table 6.1 shows the results and some of the calculations made.

Table 6.1

phenotype	**observed number (*O*)**	**expected number (*E*)**	$(O - E)^2$	$\frac{(O - E)^2}{E}$
short, black fur	25		4	0.148
long, black fur	11		4	0.444
short, white fur	8		1	0.111
long, white fur	4		1	0.333

Calculate the expected numbers and write them in the shaded column in Table 6.1. [1]

(iii) Use the formula provided and the figures in Table 6.1 to calculate the χ^2 value.

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

χ^2 = ... [1]

7 Yeasts are unicellular organisms from the kingdom Fungi. *Saccharomyces cerevisiae* is one species of yeast that can carry out either asexual reproduction by mitosis or sexual reproduction by meiosis.

Budding in *S. cerevisiae* is a process where a small daughter cell forms as a bud on the parent cell. The bud contains a copy of the parent cell nucleus and it eventually separates from the parent cell to form a new cell.

S. cerevisiae can exist in two forms: haploid cells or diploid cells.

- Haploid cells can be one of two different mating types: **a** and α.
- Haploid cells can only mate with other haploid cells of the opposite mating type.

Fig. 2.1 shows the life cycle of *S. cerevisiae* with its asexual and sexual reproductive stages.

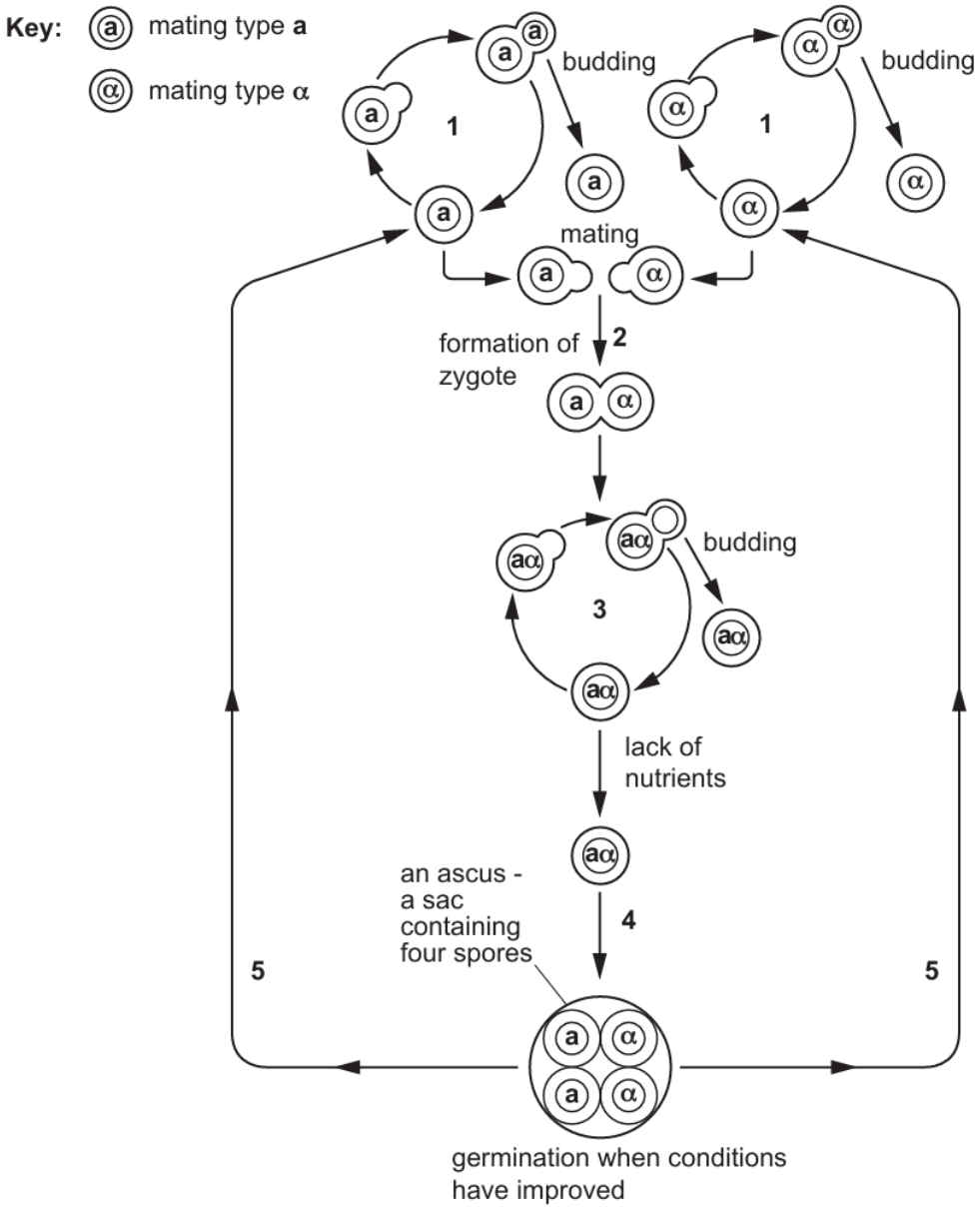


Fig. 2.1

(c) Haploid and diploid cells of *S. cerevisiae* can carry out **asexual** reproduction.

Suggest why a new harmful recessive mutation may **not** have a damaging effect on:
- an asexually reproducing population of haploid cells of *S. cerevisiae*
- an asexually reproducing population of diploid cells of *S. cerevisiae*.

.....

.....

.....

.....

.....

.....

..... [3]

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

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

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

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

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

Table 5.1

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

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

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

The value of χ^2 = 2097.836.

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

Table 5.2

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

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

.....

.....

.....

.....

.....

.....

.....

.....

.....

..... [4]

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

.....

.....

.....

.....

.....

.....

.....

.....

.....

..... [4]

9 The tiger barb, *Puntigrus tetrazona*, is a South American fish that is popular worldwide as an aquarium fish. Fig. 6.1 shows the appearance (phenotype) of a normal (wild-type) tiger barb.



Fig. 6.1

- Tiger barbs that show a wild-type phenotype are gold with black stripes.
- Tiger barbs that show an albino phenotype are gold with white stripes.
- In 2012, a fish breeder discovered a tiger barb with a new, transparent, phenotype. This fish had a transparent body and black stripes.

The fish breeder crossed the tiger barb showing the new transparent phenotype with a tiger barb showing the albino phenotype.

All the F1 offspring were wild-type. These F1 offspring were crossed with each other.

Table 6.1 shows the phenotypes obtained in the F2 generation and the number of fish showing each phenotype.

Table 6.1

F2 phenotype	number of fish
wild-type (gold with black stripes)	173
albino (gold with white stripes)	57
transparent with black stripes	58
transparent with white stripes	19

(a) (i) The fish breeder concluded that the new transparent phenotype had occurred because of a mutation.

Explain how the results in Table 6.1 support this conclusion.

.....

.....

.....

..... [2]

(ii) State the approximate whole-number ratio shown by the results in Table 6.1.

..... [1]

(iii) Explain what the results in Table 6.1 show about the genes and alleles that determine the wild-type, albino and the two different transparent phenotypes in tiger barbs.

.....

.....

.....

.....

.....

.....

.....

.....

.....

..... [4]

10 **(a)** In some varieties of domestic cats the gene for fur colour is located on the **X** chromosome. This gene has two alleles. One allele codes for black fur and the other allele codes for ginger fur. The two alleles are codominant so a heterozygous cat will have fur with patches of black and ginger colours. A cat with fur of two colours is known as a tortoiseshell.

Using appropriate symbols, construct a genetic diagram to show the results of a cross between a female tortoiseshell cat and a male ginger cat.

symbols

parent phenotypes tortoiseshell female ginger male

parent genotypes

gametes

offspring genotypes

offspring phenotypes

[5]

(b) In humans the *TYR* gene is involved in the production of a dark pigment, melanin, in some cells.

Describe how the expression of the *TYR* gene leads to the production of melanin.

.....

.....

.....

.....

.....

..... [3]

11 Two subspecies of reindeer, *Rangifer tarandus*, live in North America. Members of the different subspecies belong to the same species but have some morphological differences and are found in different geographical locations.

Fig. 5.1 shows a reindeer.



Fig. 5.1

Table 5.1 compares the features of the two North American reindeer subspecies.

Table 5.1

feature	**woodland subspecies, *R. tarandus caribou***	**barren ground subspecies, *R. tarandus groenlandicus***
habitat	southern woodland (warmer)	northern tundra (colder)
type of food	tree leaves, grass	lichens, moss
summer and winter feeding grounds overlap	yes	no
carry out long migrations	no	yes
body size	large	small
colour of fur	dark	light

(d) Outline how practical techniques could be used to test the hypothesis that migratory behaviour in reindeer has a genetic basis.

.....

.....

.....

.....

.....

.....

.....

..... [3]

12 **(a)** The fruit fly, *Drosophila melanogaster*, usually has red eyes. A gene for eye colour has four alleles: red, apricot, honey and white.

Define the terms *gene* and *allele*.

gene .....

.....

allele .....

.....
[2]

(b)
- The allele for red eyes, C^R, is dominant to the other three alleles.
- The allele for apricot eyes, C^A, is dominant to the allele for honey eyes, C^H.
- The allele for white eyes, C^W, is recessive to the other three alleles.

Construct a genetic diagram to show the genotypes, phenotypes and ratio of the offspring from a cross between a fruit fly with red eyes, C^RC^H, and a fruit fly with apricot eyes, C^AC^W.

parents phenotypes	red eyes	apricot eyes
parents genotypes	C^RC^H	C^AC^W
gametes		
offspring genotypes		
offspring phenotypes		
ratio		

[3]

(c) Describe how you would carry out a test cross **and** use it to determine the genotype of a red-eyed fruit fly.

.....

.....

.....

.....

.....

.....

..... [3]

13 The jaguar, *Panthera onca*, is a large cat that lives mainly in South America. The majority of jaguars have light brown fur with black spots, as shown in Fig. 1.1. Some jaguars have completely black fur, as shown in Fig. 1.2.

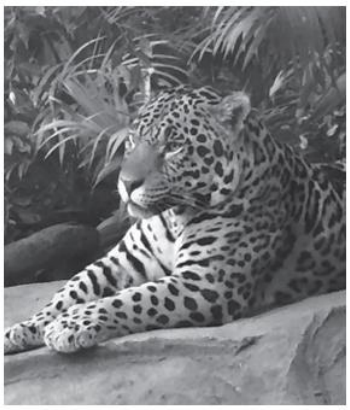


Fig. 1.1



Fig. 1.2

(a) The pigments involved in fur colour are produced as a result of biochemical pathways that take place in cells called melanocytes. These pathways are similar to those that occur in human melanocytes.

The melanocortin 1 receptor (MC1R) is located on the cell surface membrane of melanocytes and is coded for by the *MC1R* gene.

Fig. 1.3 outlines the processes that occur in jaguar melanocytes.

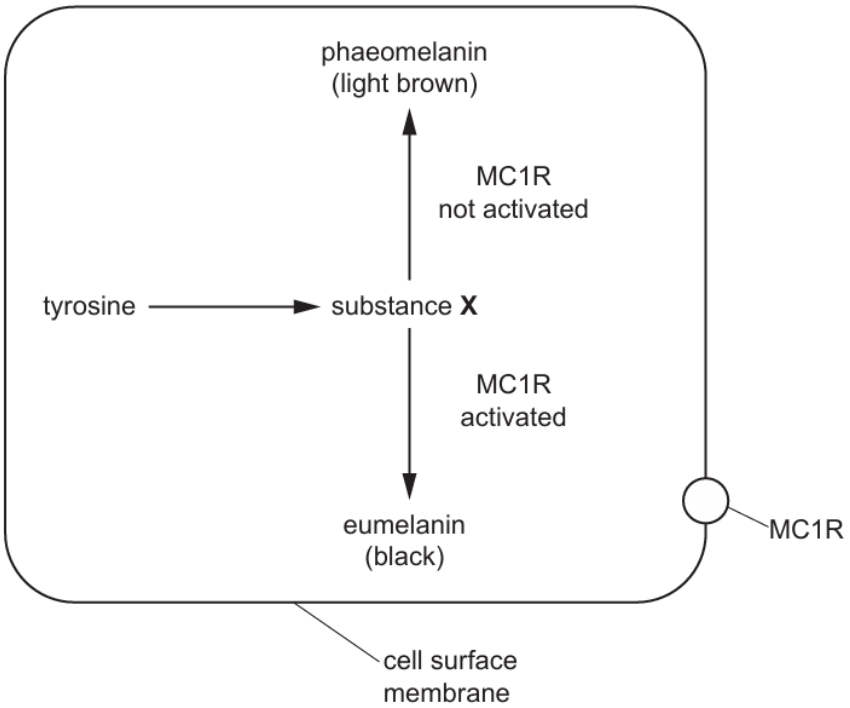


Fig. 1.3

(i) Name the substance represented by **X**.

..... [1]

(ii) When MC1R is activated a second messenger is produced in the cell.

Give an example of a second messenger.

..... [1]

(iii) Substance **X** is also produced in humans, but a mutation of the *TYR* gene can result in substance **X** not being produced.

Describe the phenotype of a person with this mutation.

.....

.....

.....

.....

..... [2]

(b) The *MC1R* gene has two alleles and is located on an autosome.

- When two jaguars with light brown fur mate all the offspring have light brown fur.
- When two jaguars with black fur mate either all the offspring will have black fur or some offspring will have black fur and some will have light brown fur.

Using symbols, construct a genetic diagram to show how two jaguars with black fur can produce some offspring with black fur and some offspring with light brown fur.

[4]

14 **(a)** The fruit fly, *Drosophila melanogaster*, feeds on sugars found in damaged fruits.

A fruit fly with normal features is described as wild type. It has red eyes and its wings are longer than its abdomen. There are mutant variations such as purple eyes or short (vestigial) wings.

Fig. 7.1 shows a wild type fruit fly and a mutant fruit fly with purple eyes and vestigial wings.



Fig. 7.1

- The genes coding for eye colour and wing length are located on the same chromosome.
- Allele **R** for red eyes is dominant to allele **r** for purple eyes.
- Allele **N** for long wings is dominant to allele **n** for vestigial wings.

Explain what is meant by the terms allele and dominant.

allele .....

dominant .....

.....

[2]

(b) A wild type fruit fly, heterozygous for both genes, was crossed with a fruit fly that was homozygous recessive for both genes.

(i) Table 7.1 is a summary of the cross.

Complete Table 7.1.

Table 7.1

	wild type parent		**double homozygous recessive parent**	
parental phenotype	..... x		....	
parental genotype	..... x			
offspring				
offspring phenotype	....	....	....	....
offspring genotype	.....	.....	.....	.....
number of offspring	1339	1195	151	154

[5]

(ii) Explain why the four offspring phenotypes are not present in a 1:1:1:1 ratio.

.....

.....

.....

.....

.....

.....

..... [3]

15 People vary in which molecules are present on the surface of their red blood cells. For example, in the ABO blood group system, people may be categorised into one of four blood groups: A, B, AB and O.

A single gene, ***I***, with three alleles, determines these blood groups. Gene ***I*** is an example of a gene with multiple alleles.

- Alleles I^A and I^B are codominant.
- Allele I^o is recessive to alleles I^A and I^B.

(a) Explain what is meant by codominant and recessive.

codominant .....

.....

recessive .....

.....

[3]

(b) Table 7.1 shows a cross between a person with blood group A and a person with blood group B.

Complete Table 7.1.

Table 7.1

parental phenotype (blood group)	A		B	
parental genotype	$I^A I^o$		$I^B I^o$	
gametes	.....	.....	.....	.....
offspring genotype	.....	.....	.....	.....
offspring phenotype (blood group)	.....	.....	.....	.....

[3]

(c) Haemophilia is a blood condition where bleeding continues for a long period of time after a blood vessel has been damaged.

A person with haemophilia has a mutation in a gene on the X chromosome, which results in the lack of a blood clotting factor.

Explain why a man who has haemophilia cannot pass the condition to his male offspring.

.....

.....

.....

.....

.....

.....

..... [3]

16 **(a)** The Labrador is a variety of domestic dog. Labradors have fur that can be brown, black or yellow.

In Labradors, *TYRP1* is one gene that codes for fur colour. This gene has two alleles, **B** and **b**.

- The dominant allele, **B**, codes for the enzyme tyrosinase that functions in the pathway to produce melanin, leading to black fur.
- The production of melanin in Labradors is very similar to the production of melanin in humans.
- The recessive allele, **b**, codes for an enzyme that results in the production of a brown form of melanin, leading to brown fur.

Outline how melanin may be produced in Labradors to produce black fur.

.....

.....

.....

.....

..... [2]

17 The summer squash plant, *Cucurbita pepo*, produces edible fruits that vary in shape.
Fig. 2.1 shows the fruits of three different varieties of squash plants.



Patty pan
(disc-shaped)



Di Nizza
(spherical)



Alfresco
(long)

Fig. 2.1

Fruit shape in squashes is controlled by two genes, **A/a** and **B/b**, that are located on different chromosomes.

- A disc-shaped fruit is produced when both dominant alleles, **A** and **B**, are present.
- A spherical fruit is produced when either allele **A** or allele **B** is present, but not if both **A** and **B** are present.
- A long fruit is produced when both allele **A** and allele **B** are absent.

(a) (i) Table 2.1 shows the possible genotypes of the Patty pan and Alfresco varieties.

Complete Table 2.1 to show the possible genotypes of the Di Nizza variety.

Table 2.1

variety	**possible genotypes**			
Patty pan (disc-shaped)	**AABB**	**AaBB**	**AABb**	**AaBb**
Di Nizza (spherical)	.....	.....	.....	.....
Alfresco (long)	**aabb**			

[1]

(ii) A gardener used pollen from a male flower of Alfresco to pollinate a female flower of Di Nizza. The gardener grew the seeds produced from this cross and found that half the offspring produced spherical fruits and half produced long fruits.

Draw **one** genetic diagram to explain this result.

parent genotypes

gametes

offspring genotypes

offspring phenotypes

[4]

(iii) The offspring show genetic variation with respect to fruit shape alleles.

Name the process that occurred during meiosis in the parents that produced this variation **and** state the stage of meiosis at which it occurred.

process .....

stage of meiosis ..... [2]

18 **(d)** Some plants are grown commercially for their flowers. Many of these plants are varieties that have short stems.

Two factors that affect the height of stems are:

- gibberellin
- the *Le*/*le* gene.

The *Le*/*le* gene has two alleles, Le and le.

(i) Suggest an advantage of growing a short-stemmed variety of a flowering plant.

.....

.....

..... [1]

(ii) Explain how the *Le*/*le* gene and gibberellin are involved in affecting the height of plant stems.

.....

.....

.....

.....

.....

.....

..... [3]

19 **(b)** Hunter syndrome is caused by a recessive allele located on the **X** chromosome.

Using appropriate symbols, draw a genetic diagram to show the probability of a normal male and a heterozygous normal female having a child with Hunter syndrome.

key

parental phenotype	normal male	×	normal female
parental genotype			
gametes			
offspring genotypes			
offspring phenotypes			

probability ..... [5]

20 The patty pan squash plant, *Cucurbita pepo*, produces edible fruits that vary in colour.

(a) The colour of the fruits is controlled by two genes, **A/a** and **B/b**, that occur on different chromosomes.

- Allele **A** produces a white fruit colour.
- Allele **a** does not produce a colour by itself but allows the colours coded by gene **B/b** to show in the phenotype.
- Allele **B** produces a yellow fruit colour.
- Allele **b** produces a green fruit colour.

In a dihybrid cross, an **AABB** plant was crossed with an **aabb** plant. All the resulting F1 plants produced white fruits.

The F1 plants were then crossed with each other to obtain the F2 generation.

(i) Complete Fig. 2.1 with the gametes produced by the F1 parents, the F2 genotypes and the F2 phenotypes.

State the ratio of fruit colours in the F2 offspring.

F1 parents: **AaBb** × **AaBb**

Fig. 2.1

ratio of fruit colours in F2 offspring .....

[4]

(ii) Test crosses were carried out on two white-fruited plants, **P** and **Q**, from the F2 generation. Each of these plants had its female flowers pollinated with pollen from a green-fruited plant.

For plant **P**, half of the offspring were white and half were yellow.
For plant **Q**, half of the offspring were white and half were green.

Deduce the genotypes of plants **P** and **Q**.

plant **P** .....

plant **Q** .....

[2]

21 Sickle cell anaemia is a non-infectious chronic disease. If not treated, sickle cell anaemia can be painful and life-threatening.

Sickle cell anaemia is caused by a base substitution mutation in the gene coding for the β-globin polypeptide of haemoglobin. This leads to a change in the primary structure of the polypeptide, as valine is present instead of glutamine. This results in abnormal sickle-shaped red blood cells, which stick together in blood vessels.

Symptoms of sickle cell anaemia include painful attacks when red blood cells block capillaries in tissues and organs.

(a) Suggest the consequences to cells when sickle-shaped red blood cells block capillaries in tissues and organs.

.....

.....

.....

..... [2]

22 Tyrosinase is an enzyme found in mammals. It is involved in the synthesis of melanin pigment. Mutations in the tyrosinase gene affect a mammal's hair colour.

Table 5.1 compares DNA sequences for codons 974–985 of:
- the normal tyrosinase gene of humans (human)
- the normal tyrosinase gene of cats that have pigmented hair (normal cat)
- the tyrosinase gene of cats that show an albino phenotype (albino cat).

The corresponding amino acid sequences of each tyrosinase are shown in the shaded rows.

Table 5.1

	974	**975**	**976**	**977**	**978**	**979**	**980**	**981**	**982**	**983**	**984**	**985**
human	CTC	CCC	TCT	TCA	GCT	GAT	GTG	GAA	TTT	TGC	CTA	AGT
normal cat	CTC	CCC	TCC	TCT	GCT	GAT	GTG	GAA	TTT	TGC	CTA	AGT
albino cat	CTC	CCT	CCT	CTG	CTG	ATG	TGG	AAT	TTT	GCC	TAA	GTC
human	Leu	Pro	Ser	Ser	Ala	Asp	Val	Glu	Phe	Cys	Leu	Ser
normal cat	Leu	Pro	Ser	Ser	Ala	Asp	Val	Glu	Phe	Cys	Leu	Ser
albino cat	Leu	Pro	Pro	Leu	Leu	Met	Trp	Asn	Phe	Ala	STOP	–

(a) (i) A silent mutation involves a base substitution that does not result in an amino acid change.

Use Table 5.1 to identify, with reasons, a silent mutation distinguishing humans from normal cats with pigmented hair.

.....

.....

.....

.....

.....

.....

..... [2]

(ii) State the changes that resulted in the premature STOP codon in the albino cat DNA sequence.

.....

.....

.....

.....

..... [2]

(iii) Explain why albino cats, homozygous for the mutation that resulted in the premature STOP codon, do **not** produce melanin.

.....

.....

.....

.....

..... [2]

(iv) Bioinformatics was used to compare the whole sequence of the tyrosinase genes of humans and cats.

Explain why bioinformatics was used to compare these gene sequences **and** suggest a conclusion that could be made from the percentage similarity data obtained.

.....

.....

.....

.....

.....

..... [2]

(b) Siamese cats have a temperature-sensitive tyrosinase that only functions in the cooler areas of the skin. This means they only produce a small quantity of melanin pigment. Melanin is mainly on their ears, face, paws and tail.

Fig. 5.1 shows a Siamese cat.



Fig. 5.1

The Siamese allele of tyrosinase (t^s) is recessive to the normal allele that causes full pigmentation all over the body (**T**) but is dominant to the albino allele (t^a).

Draw a genetic diagram to show how a cross between an albino cat and a fully pigmented cat can result in offspring that include kittens with Siamese colouring.

parent phenotypes: albino × fully pigmented

parent genotypes:

gametes:

F1 genotypes:

F1 phenotypes:

ratio:

[3]

23 There are a number of mutations affecting the production of fetal haemoglobin, HbF, and normal adult haemoglobin, HbA.

- The Hb^A allele codes for the normal β-globin polypeptide of haemoglobin.
- The Hb^S allele, caused by a base substitution mutation, codes for an abnormal β-globin polypeptide.
- The base substitution results in the amino acid glutamine, which has a polar R group, to be replaced by valine, which has a non-polar R group, in the polypeptide.

The abnormal haemoglobin molecules (HbS) form fibres in low partial pressures of oxygen (pO_2). The fibres cause red blood cells to become sickle shaped and the cells can block blood capillaries.

Individuals with adult haemoglobin molecules that are all abnormal (HbS) have sickle cell anaemia. This is a painful chronic condition that can be life-threatening.

(a) Explain why this mutation causes the HbS to form fibres.

.....

.....

.....

..... [2]

24 Fig. 5.1 shows a cat displaying the recessive phenotype for two unlinked genes.

- **H/h** controls hair length. The allele for short hair is dominant to the allele for long hair.
- **R/r** controls coat pattern. The allele for 'full colour', with pigment on all parts of the body, is dominant to the allele for 'pointed', where the pigment is restricted to the ears, face, paws and tail.



Fig. 5.1

(a) Draw a genetic diagram to predict the offspring genotypes and phenotypes when the cat in Fig. 5.1 is crossed with a cat that is heterozygous for the hair length gene and heterozygous for the coat pattern gene.

parent phenotypes:

parent genotypes:

gametes:

F1 genotypes:

F1 phenotypes:

[5]

(b) Scientists isolated two sections of DNA thought to correspond with the allele for full colour (**R**) and the allele for pointed (**r**).

Sequencing these two DNA sections showed that the DNA sequence for full colour, **R**, had a restriction site for the restriction enzyme *Hpa*II. This restriction site did **not** occur in the DNA sequence **r** because of a single nucleotide substitution.

The scientists then carried out an analysis of three generations of cats. Each cat was assessed for three features:

- coat pattern, full colour or pointed
- the presence or absence of the *Hpa*II restriction site
- the pair of alleles present at a variable marker locus thought to lie close to the **R/r** locus. The marker locus (gene) has seven different alleles designated as 1, 2, 3, 4, 5, 6 and 7.

Fig. 5.2 shows the relationships of these cats and the results of the assessment.

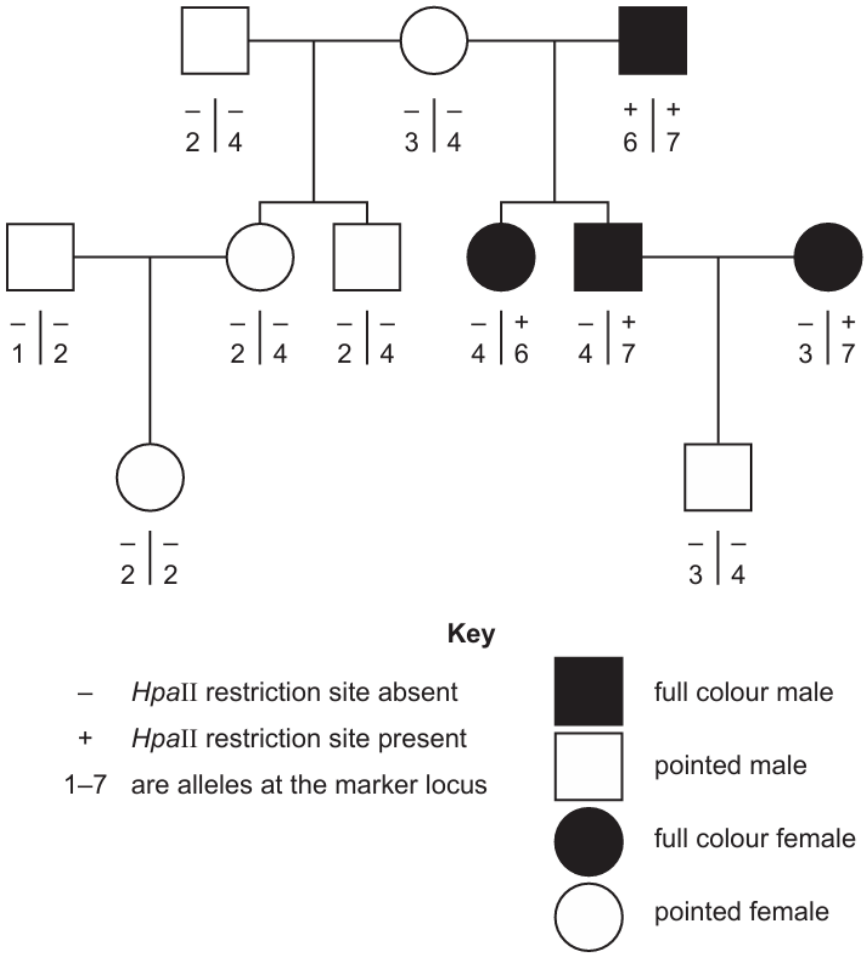


Fig. 5.2

Identify evidence from Fig. 5.2 to support these statements:

(i) the pointed phenotype is due to a recessive allele

.....

.....

..... [1]

(ii) the **R/r** gene is located on an autosome

.....

.....

.....

.....

..... [2]

(iii) the marker locus and **R/r** are closely linked.

.....

.....

.....

.....

..... [2]

25 **(b)** Flower colour is important in sexual reproduction of insect-pollinated plants.

In the rosy periwinkle, *Catharanthus roseus*, flower colour is controlled by three genes, **R/r**, **D/d** and **P/p**, which interact together to control flower colour.

Fig. 4.1 is a drawing of a rosy periwinkle.

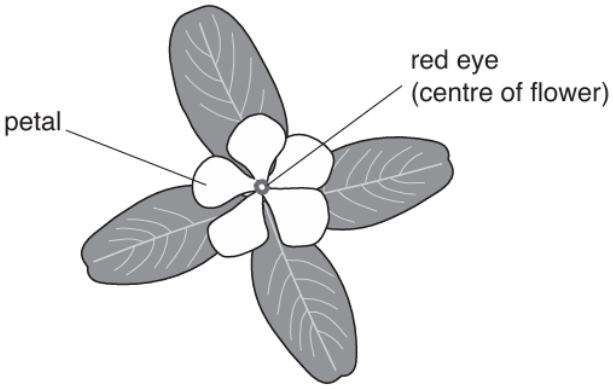


Fig. 4.1

The presence of the **R** allele results in a red pigment in the centre of the flower (red eye).

The **D** allele and the **P** allele are only expressed when the **R** allele is present.

- When the **D** allele and the **R** allele are present, the flower has dark pink petals with a red eye.
- When the **P** allele and the **R** allele are present, the flower has pale pink petals with a red eye.
- When the **D** allele, the **P** allele and the **R** allele are all present, the flower has dark pink petals with a red eye.
- The recessive alleles **r**, **d** and **p** result in no pigments being produced and the flower has white petals and no red eye.

(i) Deduce the phenotypes of these rosy periwinkle genotypes.

RR dd PP .....

Rr Dd Pp .....

rr Dd Pp .....

RR dd pp ..... [4]

(ii) The pigments causing flower colour in the rosy periwinkle are formed by a biosynthetic pathway.

The **R** allele mutated to produce the **r** allele.

The **r** allele codes for a non-functional protein.

Explain how the mutation that changes **R** to **r** results in no red pigment being synthesised in the flower of rosy periwinkle.

.....

.....

.....

.....

.....

.....

.....

.....

.....

..... [4]

26 Oculocutaneous albinism (OCA) is a type of albinism. There are many different forms of OCA.

OCA1A is one form of OCA, caused by a recessive mutation in the autosomal gene, *TYR*, coding for the enzyme tyrosinase. This enzyme is involved in the biosynthetic pathway that results in the production of melanin, the pigment responsible for the colour of hair, skin and eyes.

A person with OCA1A has white hair, very pale skin and pink eye colour.

(a) Draw a genetic diagram to show the probability of a child having OCA1A, if both parents are carriers.

Use the symbols **A** and **a** for the alleles.

parental genotypes

gametes

offspring genotypes

offspring phenotypes

probability

[4]

27 Some neurones in the brain produce a neurotransmitter known as dopamine. Parkinson's disease occurs when the neurones that produce dopamine die. A person with the disease may experience difficulty in coordinating movement, often seen as tremors (shaking) in different parts of the body.

Parkinson's disease typically occurs in people older than 55 years. Younger people with these symptoms are said to have early onset Parkinson's disease (EOPD).

Recessive mutations in a gene known as *PINK1*, located on chromosome 1, an autosome, are believed to be one cause of EOPD. A person with this form of EOPD has a homozygous recessive genotype.

(a) Draw a genetic diagram of a cross between two individuals who are heterozygous at the *PINK1* gene locus.

key to symbols used for alleles

.....

.....

parental genotypes

gametes

offspring genotypes

ratio of offspring phenotypes

[4]

(c) One rare, dominant mutation of the *PINK1* gene codes for a product that inhibits the normal protein kinase.

Explain how this mutation causes EOPD in a heterozygote.

.....

.....

.....

.....

..... [2]

28 **(a)** In the sweet pea plant, *Lathyrus odoratus*, one gene codes for flower colour and one gene codes for pollen grain shape.
Flower colour is either purple or red. Pollen grain shape is either long or round.
The inheritance of these genes is an example of **autosomal linkage**.

- The allele **F** for purple flowers is dominant over the allele **f** for red flowers.
- The allele **G** for long pollen grains is dominant over allele **g** for round pollen grains.

Explain the meaning of the term *autosomal linkage*.

.....

.....

.....

.....[2]

(b) A dihybrid cross was carried out between homozygous dominant and homozygous recessive sweet pea plant parents to produce the F1 generation.

The offspring from the F1 generation were crossed to produce the F2 generation.

(i) Draw a genetic diagram to show a dihybrid cross between two offspring from the F1 generation.

Assume that these genes are closely linked and that there are **no** crossing over events.

[4]

(ii) The actual results of the dihybrid cross are shown in Table 2.1.

Table 2.1

phenotypes of F2 offspring	number of individuals
purple flowers, long pollen grains	284
purple flowers, round pollen grains	21
red flowers, long pollen grains	21
red flowers, round pollen grains	55

State how the results support the fact that this is an example of autosomal linkage.

.....

.....

.....[1]

(c) **(i)** In a test cross, an individual of **known** genotype is crossed with an individual that has a dominant phenotype but unknown genotype.

State the genotype of the **known** individual in a test cross.

.....[1]

(ii) A test cross was carried out with sweet pea plants known to be heterozygous for both flower colour and pollen grain shape.

The results of the test cross are shown in Table 2.2.

Table 2.2

phenotypes of offspring of test cross	number of individuals
purple flowers, long pollen grains	215
purple flowers, round pollen grains	30
red flowers, long pollen grains	32
red flowers, round pollen grains	210

The result of a test cross can be used to determine a crossover value (COV).
A crossover value is the percentage of the total number of offspring showing recombination.

The crossover value (COV) can be calculated using the formula shown in Fig. 2.1.

$$\text{COV} = \frac{\text{number of recombinants}}{\text{total number of individuals}} \times 100$$

Fig. 2.1

Calculate the COV from the results shown in Table 2.2.

COV = ...% [1]

(iii) Suggest what information about the relative distance between the linked genes can be gained from crossover values.

.....

.....[1]

29 Domestic goats, *Capra hircus*, show a wide range of coat patterns and colours. One gene involved in coat colour and pattern has multiple alleles. Four of these alleles are:

- **A**, the allele for white, is dominant to all others
- $\mathbf{A^b}$, the allele for badgerface (stripes on face) and $\mathbf{A^g}$, the allele for grey, are codominant
- **a**, the allele for black, is recessive to all others.

(a) State **all** the possible genotypes of:

a grey badgerface goat .....

a white goat. .....

[2]

(b) A cross between a black goat and a white goat produced a white goat. This white offspring was crossed with a grey goat. The genotype of the grey goat was not known.

Use genetic diagrams to show **all** the possible offspring genotypes and phenotypes that could result from the cross between the white offspring and the grey goat.

[4]

(c) When goat skin cells produce black melanin pigment these events occur:

- Melanocyte stimulating hormone (MSH) is secreted into the bloodstream.
- MSH binds to the melanocortin 1 receptor on the cell surface membranes of skin cells.
- This causes a rise in cyclic AMP within the cell.
- The enzyme tyrosinase is activated and begins melanin production.

The white allele **A** codes for a protein that binds irreversibly to the melanocortin 1 receptor. No rise in cyclic AMP occurs, even when MSH is present in the blood.

Explain the appearance of goats with genotype **aa**.

.....

.....

.....

.....

.....

.....[2]

30 In the fruit fly, *Drosophila melanogaster*, two different genes control body colour and eye colour.

- **G**/**g** are alleles of the body colour gene.
- **G** results in grey body, **g** results in black body.

- **R**/**r** are alleles of the eye colour gene.
- **R** results in red eyes, **r** results in brown eyes.

Each gene is autosomal.

A dihybrid cross was carried out using a fly with a grey body and red eyes crossed with a fly with a black body and brown eyes. Both parents were homozygous for both genes. The offspring from the F1 generation were crossed to obtain the F2 offspring.

(a) A statistical test showed that the results of the cross were significantly different from those expected.

State the name of the statistical test used **and** state the expected phenotypic ratio for the F2 generation.

statistical test .....

expected ratio .....

[2]

(b) A test cross can be carried out in order to identify flies from an F2 generation that are heterozygous for both genes.

Draw a genetic diagram to show how a test cross between a heterozygous grey-bodied, red-eyed F2 fly and a fly with a black body and brown eyes can produce four different offspring phenotypes.

Use the symbols **G**/**g** and **R**/**r**.

[4]

(c) The results of the test cross in **(b)** are shown in Table 2.1. These results are significantly different from the expected results.

Table 2.1

phenotypes of offspring of test cross	number of individuals
grey body, red eyes	123
grey body, brown eyes	7
black body, red eyes	6
black body, brown eyes	132

Describe how these results are different from the expected results **and** explain why they are different.

.....

.....

.....

.....

.....

.....

.....

.....

.....

..... [5]