Inheritance Pack 2

These practice questions can be used by students and teachers and is

Suitable for AQA A Level 7402 Biology Topic Question

Level: AQA A LEVEL 7402

Subject: Biology

Exam Board: AQA A Level 7402

Topic: Inheritance Pack 2

1 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 _____ _____

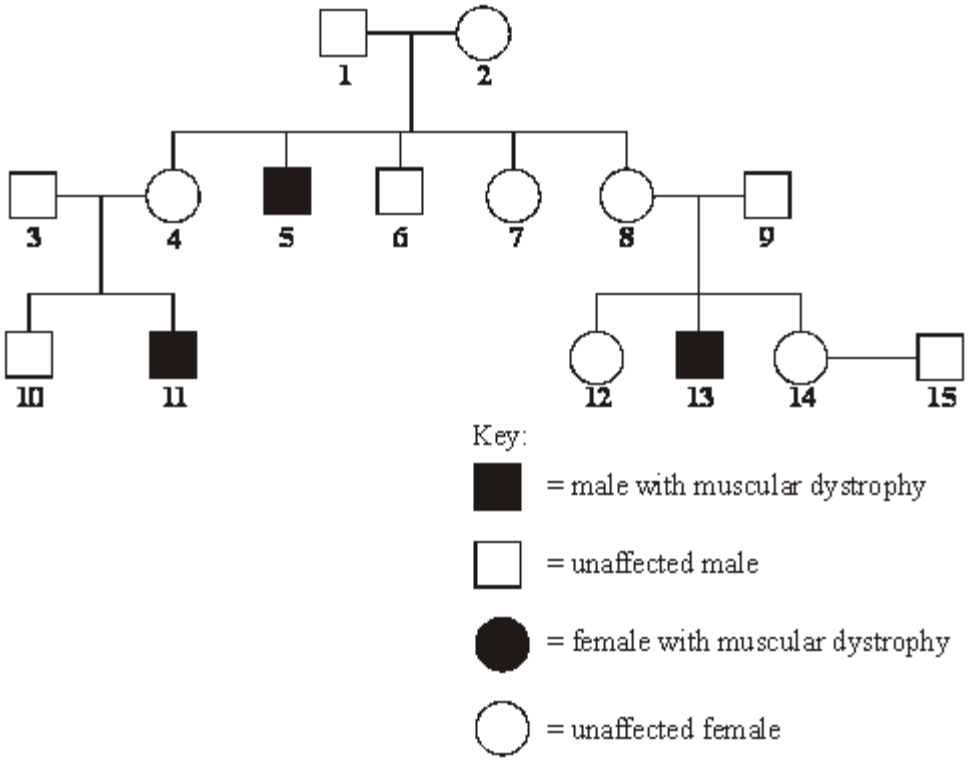
Gamete genotypes _____ _____

Offspring genotypes _____

Offspring phenotypes _____

(Total 4 marks)

2 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)

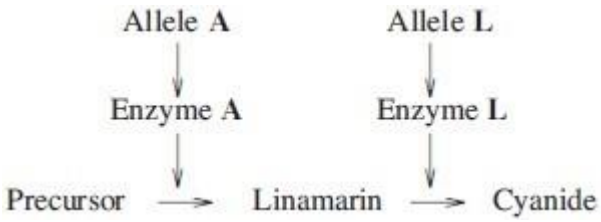
(c) A blood test shows that person **14** is a carrier of muscular dystrophy. Person **15** has recently married person **14** but as yet they have had no children. What is the probability that their first child will be a male who develops muscular dystrophy?

_____

(1)

(Total 5 marks)

3 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 proportionof 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 x^2 test was carried out on the results.

(i) Suggest the null hypothesis that was tested.

_____

_____

(1)

(ii) x^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.

(Extra space) _____

(4)

(Total 15 marks)

4 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 pairof codominant alleles, $\mathbf{C^M}$ and $\mathbf{C^N}$.

(i) What is meant by *codominant*?

_____

_____

(1)

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

Answer _____

(2)

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

Answer _____

(3)

(Total 10 marks)

5 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 thegene 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 aredominated by very tall grass plants. Snowy tigers are less successful hunters. Suggest why.

_____

_____

_____

_____

(2)

(Total 7 marks)

One form of baldness in humans is controlled by two alleles, **B** and **b**, of a single gene. This gene

6

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 $\mathbf{X^A}$ leads to normal colour vision and the recessive allele $\mathbf{X^a}$ 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 hasnormal 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)

7 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.

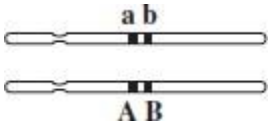
A farmer crossed a male chicken with the genotype **AaBb** with a female chicken that had a single comb and produced white eggs.

(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 eventuallyproduce blue eggs. Explain how.

_____

_____

_____

_____

(Extra space) _____

_____

(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.

_____

_____

_____

_____

(Extra space) _____

_____

(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 slowfeather 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^fY
B	$X_F X_f$	X^fY
C	$X_f X_f$	X^FY
D	$X_F X_f$	X^FY

Answer _____

Explanation _____

_____

_____

_____

(Extra space) _____

_____

(3)

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

_____

_____

_____

_____

_____

_____

(Extra space) _____

_____

(3)

(Total 14 marks)

8 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.

Parental phenotypes	Wiry-haired male	Non-wiry, long-haired female
Parental genotypes	**HhDd**	**hhdd**
Gametes		
Offspring genotypes		
Offspring phenotypes		
Ratio of offspring phenotypes		

(3)

(Total 6 marks)

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

_____

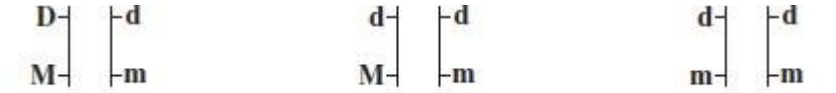
_____

_____

_____

(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.

Parental phenotypes Abnormal male Normal female

Parental genotypes

Gametes _____ _____

Offspring genotype(s) _____

Offspring phenotype(s) _____

(3)

(Total 5 marks)

10 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

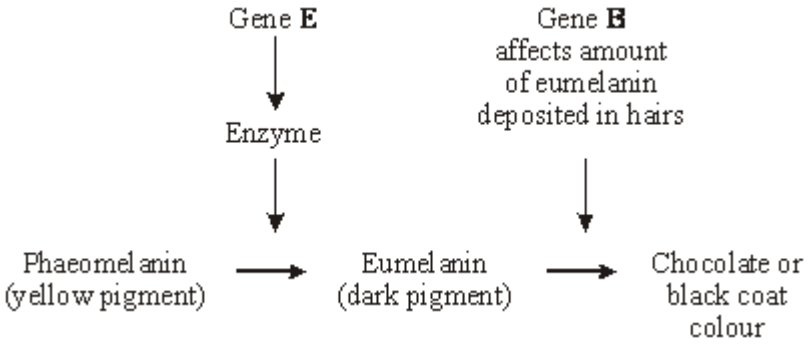
Offspring genotypes

Offspring phenotypes

Ratio of offspring phenotypes

(3)

(c) The yellow coat colour of Labrador dogs is due to the presence of the pigmentphaeomelanin 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)

11 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.

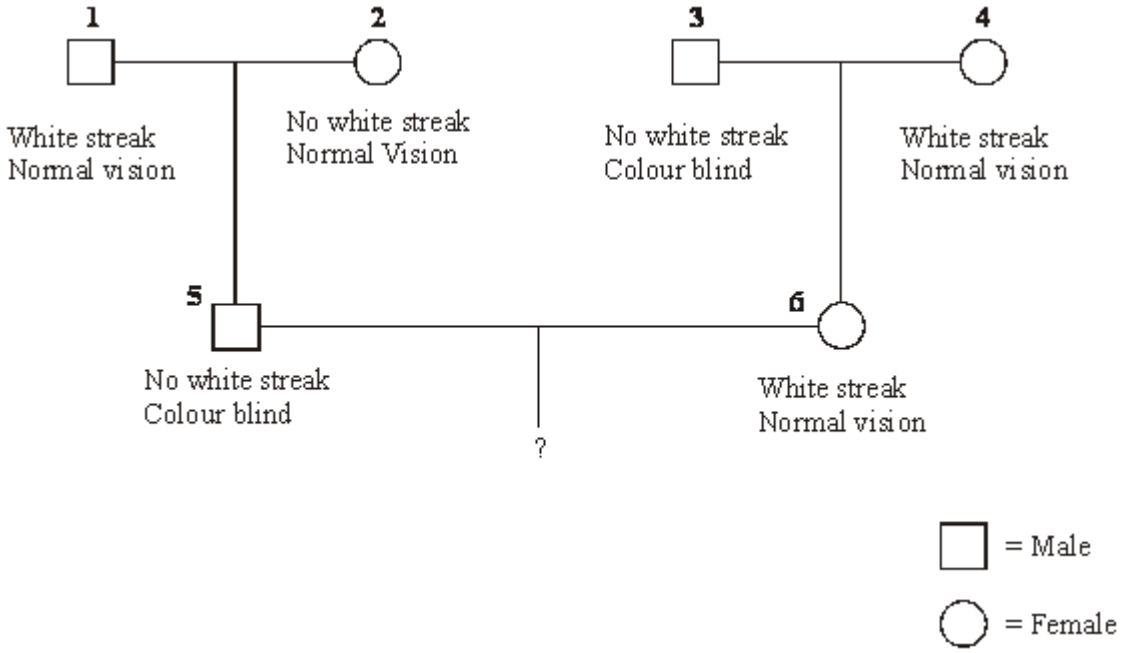
_____

_____

_____

(2)

(b) The diagram shows a family tree in which some of the individuals have colour blindness orhave 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)

12 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 $\mathbf{C^R}$ (red) and $\mathbf{C^W}$ (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.Compete 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 thesemen 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 ofthe other kind.

_____

_____

_____

_____

(2)

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

_____

_____

(1)

(Total 9 marks)

S **13** 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 tobe determined.

_____

_____

_____

_____

(2)

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

_____

_____

_____

_____

_____

_____

(3)

(Total 5 marks)

14 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, legsand tail.

_____

_____

(2)

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

_____

_____

_____

_____

(2)

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

_____

_____

_____

_____

(2)

(Total 9 marks)

15 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 unknowngenotype. 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)

16 (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
Genotypes	_____or_____	_____	_____	_____
Gamete genotype	_____or_____	_____	_____	_____

	Father (has antigen G)	Mother
Genotypes	_____	_____
Gamete genotypes	_____	_____

	Daughter (has antigen G)
Genotype	_____

(i) One of the grandmothers has two possible genotypes. Write these on the geneticdiagram, using the symbol $\mathbf{X^G}$ to show the presence of the allele for antigen G on the X chromosome, and $\mathbf{X^g}$ 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 inheritingantigen 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)

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

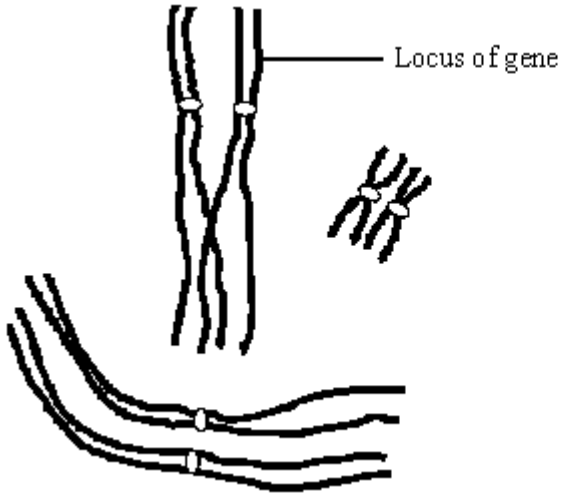


Figure 1

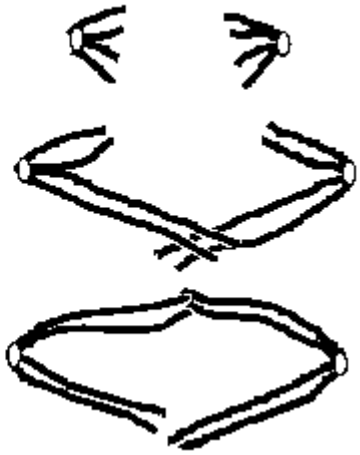


Figure 2

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

_____

(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.

(1)

(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 different pairs of homologous chromosomes. Show your working.

_____

(2)

(Total 8 marks)

18 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 $\mathbf{X^g\ Y}$ 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 phenotypesexpected in the offspring of this cross.

Parents	Male	Tortoiseshell female
Parental genotypes	**X^g Y**	_____

P*arental 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	_____	**X^gX^g Dd**

genotypes

Parental gametes

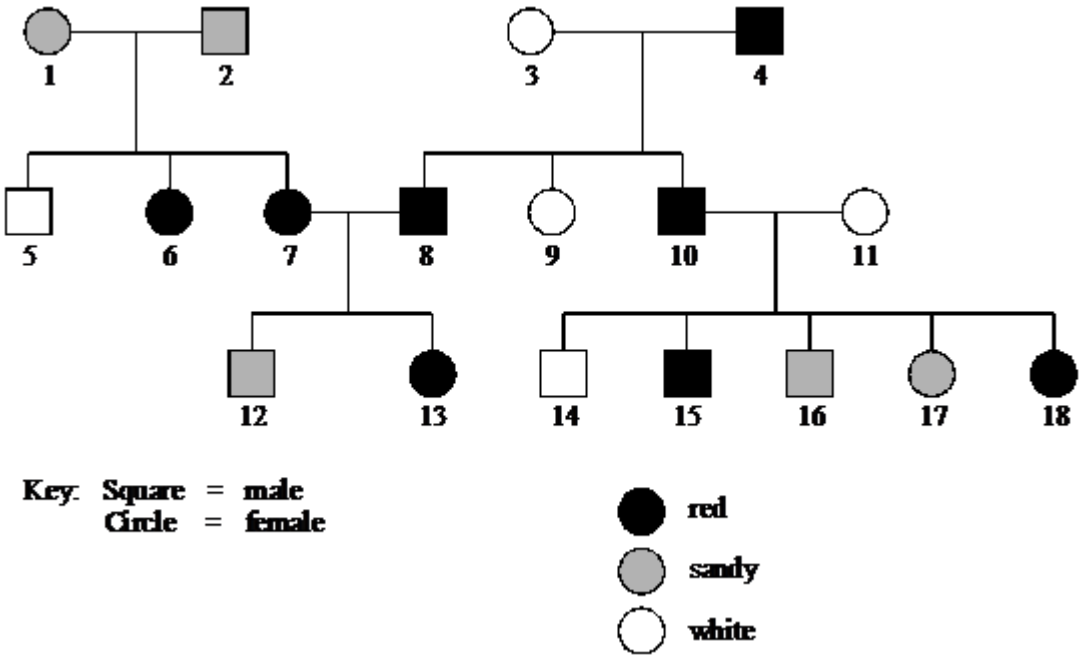
Male kitten genotypes

Male kitten colours

(3)

(Total 9 marks)

19 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.

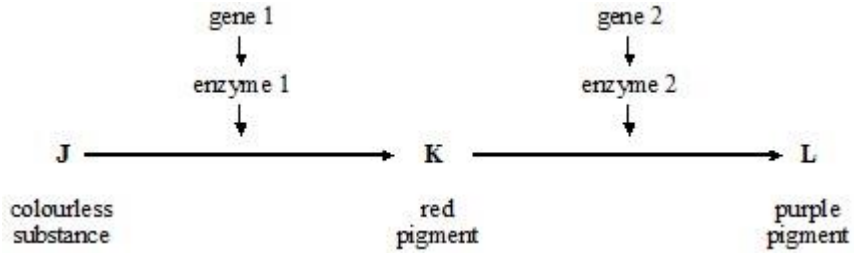
Individual 18 Other parent

Parental genotypes		**Aabb**
Parental gametes		
Offspring genotypes		
Offspring phenotypes		
Expected ratio of offspring phenotypes		

(4)

(Total 11 marks)

20 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.

_____

_____

_____

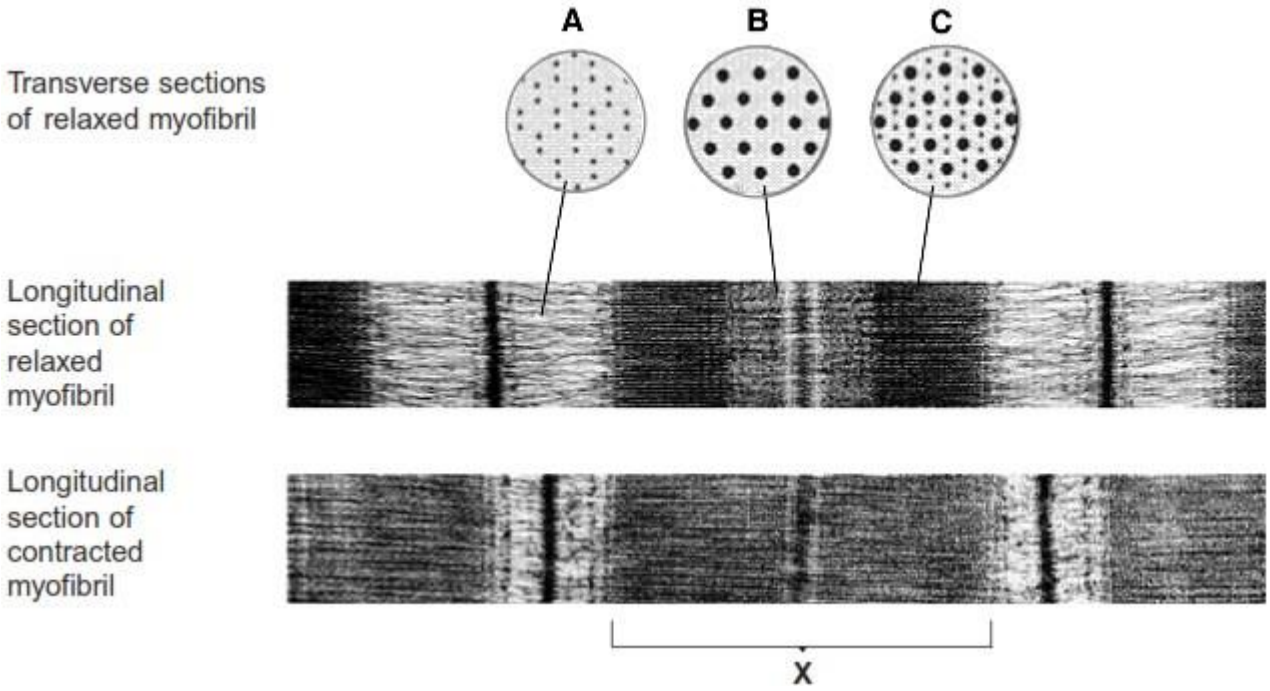
_____

_____

(2)
(Total 15 marks)

21 **Figure 1** shows sections through relaxed and contracted myofibrils of a skeletal muscle. The transverse sections are diagrams. The longitudinal sections are electron micrographs.

Figure 1



(a) (i) The electron micrographs are magnified 40 000 times.
Calculate the length of band **X** in micrometres.
Show your working.

Length of band **X** = _____ µm

(2)

(ii) Explain the difference in appearance between transverse sections **A** and **C** in **Figure 1**.

_____

_____

(1)

(b) Explain what leads to the differences in appearance between the relaxed myofibril and thecontracted myofibril.

_____

_____

_____

_____

_____

_____

_____

_____

(*Extra space*) _____

_____

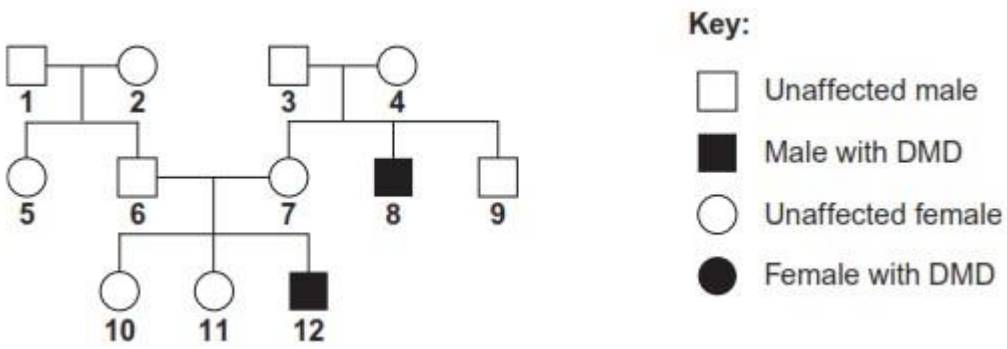
_____

_____

(4)

(c) Duchenne muscular dystrophy (DMD) is a condition caused by the recessive allele of asex-linked gene. A couple have a son with DMD. They want to know the probability that they could produce another child with DMD. They consulted a genetic counsellor who produced a diagram showing the inheritance of DMD in this family. This is shown in **Figure 2**.

Figure 2



The couple who sought genetic counselling are persons **6** and **7**.

(i) Give the evidence to show that DMD is caused by a recessive allele.

_____

_____

(1)

(ii) Give the numbers of **two** people in **Figure 2** who are definitely carriers of muscular dystrophy.

_____

(1)

(iii) Complete the genetic diagram to find the probability that the next child of couple **6** and **7** will be a son with muscular dystrophy. Use the following symbols:

$\mathbf{X}^{D}$ = normal X chromosome

$\mathbf{X}^{d}$ = X chromosome carrying the allele for muscular dystrophy

Y = normal Y chromosome

	6	**7**
Parental phenotypes	Unaffected	Unaffected
Parental genotypes	_____	_____
Gametes	_____	_____

Offspring genotypes _____

Offspring phenotypes _____

Probability of having a son with DMD _____

(4)

(d) DMD is caused by a deletion mutation in the gene for a muscle protein called dystrophin. Adeletion is where part of the DNA sequence of a gene is lost. People in different families may inherit mutations in different regions of this gene.

Scientists isolated the dystrophin gene from DNA samples taken from children **10**, **11** and **12**. They cut the gene into fragments using an enzyme. The scientists then used two DNA probes to identify the presence or absence of two of these fragments, called **F** and **G**. This allowed them to find the number of copies of each fragment in the DNA of a single cell from each child.

The table shows their results.

Child	**Number of copies of gene fragment per cell**	
	F	**G**
10 (unaffected girl)	2	1
11 (unaffected girl)	2	2
12 (boy with DMD)	1	0

(i) The number of copies of gene fragments **F** and **G** shows that person **12** has DMD. Explain how.

_____

_____

(1)

(ii) The number of copies of gene fragments **F** and **G** shows that person **12** is male. Explain how.

_____

_____

_____

_____

(2)

(iii) The genetic counsellor examined the scientists' results. He concluded that person **10** is a carrier of DMD but her sister, **11**, is not.

Describe and explain the evidence for this in the table.

_____

_____

_____

_____

_____

_____

(*Extra space*) _____

_____

_____

(3)

(e) Person **12** took part in a trial of a new technique to help people with DMD.

Doctors took muscle cells from person **12**'s father and grew them in tissue culture.

They suspended samples of the cultured cells in salt solution and injected them into a muscle in person **12**'s left leg. They injected an equal volume of salt solution into the corresponding muscle in his right leg. Person **12** was given drugs to suppress his immune system throughout the trial.

Four weeks later, the doctors removed a muscle sample from near the injection site in each leg. They treated these samples with fluorescent antibodies. These antibodies were specific for the polypeptide coded for by gene fragment **G** of the dystrophin gene.

The results are shown in the table.

Location and treatment	Percentage of muscle fibres labelled with antibody
Left leg - injected with cultured cells suspended in salt solution	6.8
Right leg - injected with salt solution	0.0

(i) Why was it necessary to treat person **12** with drugs to suppress his immune system?

_____

_____

(1)

(ii) Explain why salt solution was injected into one leg and cultured cells suspended insalt solution into the other.

_____

_____

_____

(1)

(iii) This technique is at an early stage in its development. The doctors suggested thatfurther investigations need to be carried out to assess its usefulness for treating people with DMD.

Explain why they made this suggestion.

_____

_____

_____

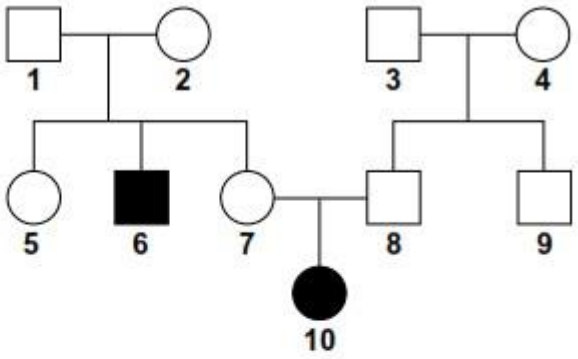
_____

(*Extra space*) _____

(4)

(Total 25 marks)

22 The diagram shows the inheritance of cystic fibrosis in one family.



Key:
- = Male with cystic fibrosis
- = Unaffected male
- = Female with cystic fibrosis
- = Unaffected female

(a) Cystic fibrosis is caused by a recessive allele.
Explain the evidence for this given in the diagram.

(2)

(b) Couple **7** and **8** decide to have another child.
What is the probability that this child will be a girl with cystic fibrosis?
Complete the genetic diagram to explain your answer.
Use the symbols **N** for the dominant allele and **n** for the recessive allele.

	7	**8**
Parental phenotypes	Unaffected	Unaffected
Parental genotypes	_____	_____
Genotypes of gametes	_____	_____

Offspring genotypes _____

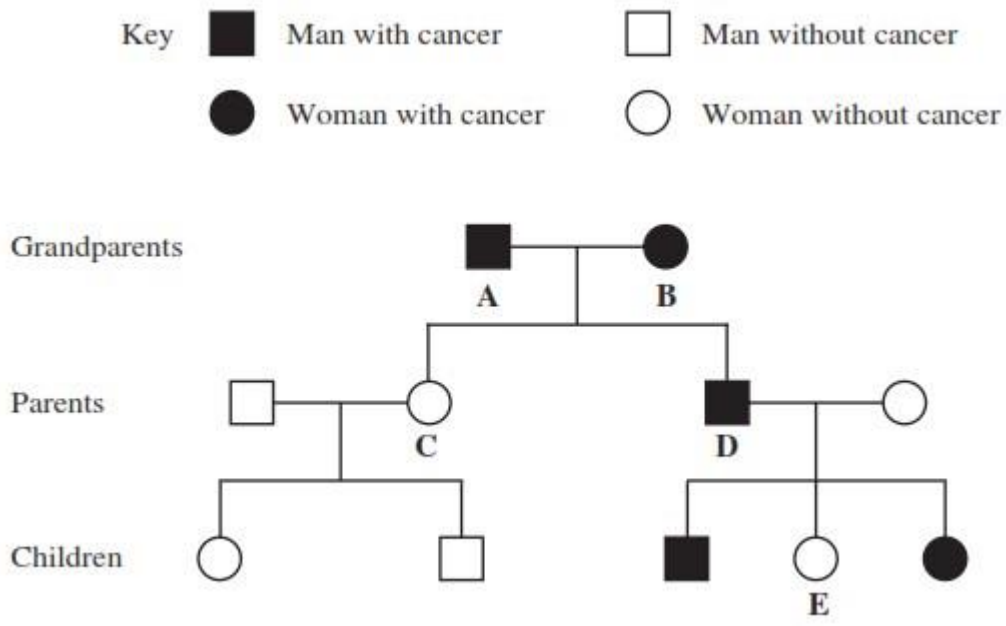
Offspring phenotypes _____

Probability of girl with cystic fibrosis _____

(4)

(Total 6 marks)

23 Li-Fraumeni syndrome is a rare inherited condition. It makes someone much more likely to develop cancer at an early age. The diagram shows part of the family history of a family affected by Li-Fraumeni syndrome. Li-Fraumeni syndrome is caused by the dominant allele of a gene. The gene is not sex-linked.



The grandparents, **A** and **B**, had two children, girl **C** and boy **D**. Explain how the phenotypes of these children provide evidence that Li-Fraumeni syndrome is

(a) caused by a dominant allele

(2)

(b) **not** sex-linked.

(2)

(c) This family's history of cancer was investigated when person **E** asked for genetic counselling. At the time she was 25 years old. What advice could a genetic counsellor give her about her probability of developing cancer?

(2)

(d) Li-Fraumeni syndrome is caused by a mutation affecting a tumour suppressor gene calledTP53. This gene codes for a protein that initiates the death of cells where damaged DNA cannot be repaired. The mutated TP53 gene leads to the production of a non-functional protein. Suggest how the non-functional protein may lead to cancer.

_____

(Extra space) _____

_____

_____

(3)

(Total 9 marks)

Mark schemes

1

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

2 **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^DX^d AND X^DX^D

8 = X^DX^d;;

All 4 correct = 2 marks
2 or 3 correct = 1 mark

max 2

(c) ¼ / 0.25 / 25% / 1:3 / 1 in 4; (*NOT* '1:4')

1

[5

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

3

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'] [Cancel: if reference to crossing-over]

2

(d) 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

(e) (i) There was no (significant) difference in damage between cyanogenic andacyanogenic / 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

(f) 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] (a) Gg / suitable equivalent;

4

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]

(a) Only expressed in the homozygote / not expressed in the heterozygote / not expressed if

5 dominant present;

1

(b) Tt Aa × tt aa ;

TA Ta tA ta ta ;

	TA	Ta	tA	ta
ta	TtAa	Ttaa	ttAa	ttaa
	Orange striped	Orange unstriped	White striped	White 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 <u>MUST</u> 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] (a) (i) BBX^AY, BbX^AY;

6

1

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

1

(b) *parental genotypes* − BbX^AY x $BbX^A X^a$;

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] (a) aabb;

7

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] (a) hhDD, hhDd;

8

(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]**

9

(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)

Gamete genotype D | d / M | m;

Offspring genotype D | d / M | m;

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

3

[5] (a) (i) black;

10

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] (a) males are XY and females XX / males have one X chromosome and females two X

11 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, HH$X^b X^b$, 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] (a) is always expressed(in the phenotype) / produces (functional) proteins;

12

1

(b) codominance;

1

(c) *Parental geneotypes* - hhC^{R}C^w , HhC^{w}C^w ;

Gametes- hC^R hC^w HC^w hC^w

Offspring geneotypes - HhC^{R}C^w , hhC^{w}C^w , HhC^{w}C^w , hhC^{w}C^w ;
Offspring pheneotypes - 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] (i) female XX, male XY;

13

Y <u>shorter / smaller</u> than X;

2

(ii) haemophilia is a recessive allele; defective allele (gene) present on X, missing from Y; male 0.5(50% / ½) probability of haemophilia; female 0 / no chance;
(0.25(25% / ¼) first baby having haemophilia);

or

X_HX_h X_HY;
$X^HX^H + X^HX^h + X^HY + X^h Y$;
X^hY is a sufferer

3 max

[5]

(a) mutations;

14 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 doesnot 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] (a) epistasis;

15

one gene influences the expression of another / description using example in question;

2

(b) aaDD, aa Dd (or DDaa, Ddaa);

1

(c) (i) AaDd (or DdAa);

1

(ii) aadd, Aadd (or ddaa, ddAa);

1

(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

[7]

16 (a) (i) paternal grandmother: $\mathbf{X^GX^G}$ or $\mathbf{X^GX^g}$

1

(ii) grandparent genotypes: [$\mathbf{X^gY}$] [$\mathbf{X^gX^g}$] [$\mathbf{X^gY}$]; gametes: [$\mathbf{X^G}$ and $\mathbf{X^g}$, or $\mathbf{X^G}$ only] [$\mathbf{X^g}$ and **Y**] [$\mathbf{X^g}$] [$\mathbf{X^g}$ and **Y**]; parents genotypes: [$\mathbf{X^GY}$] [$\mathbf{X^gX^g}$] gametes: [$\mathbf{X^G}$ and **Y**] [$\mathbf{X^g}$] daughter: [$\mathbf{X^GX^g}$];

(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 $\mathbf{X^G}$;

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] (a) 6;

17 **1**

(i) chromosomes are arranged in (homologous) pairs / bivalents;crossing over / chiasma present / exchange of genetic information; bivalents arranged independently;

2 max

(ii) separation / spliting / 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

[8]

18

(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) $\mathbf{X^GX^g}$;

(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
($\mathbf{X^g}$ and **Y** from male, $\mathbf{X^G}$ and $\mathbf{X^g}$ from female);
correct offspring genotypes ($\mathbf{X^gX^g}$, $\mathbf{X^GX^g}$, $\mathbf{X^GY}$, $\mathbf{X^gY}$);
correct link of offspring genotypes with phenotypes;
$\mathbf{X^gX^g}$ black female
$\mathbf{X^GX^g}$ tortoiseshell female
$\mathbf{X^GY}$ ginger male
$\mathbf{X^gY}$ black male

(correct gametes, offspring genotypes and link with phenotypes based on incorrect parent genotype = 3 marks)

3

(c) $\mathbf{X^GY\ dd}$; correct male kitten genotypes ($\mathbf{X^gY\ Dd}$ and $\mathbf{X^gY\ dd}$); correct link of kitten genotypes with phenotypes;

(ignore female kittens)

$\mathbf{X^gY\ Dd}$ black
$\mathbf{X^gY\ dd}$ grey

(correct kitten genotypes and phenotypes based on incorrect parent genotype = 2 marks)

3

[9]

19

(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)*	**AaBb;**	*No mark for this*
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] (a) (Gene 1) allele A makes enzyme converting J to K / colourless to red;

20

Allele a produces no / non-functional <u>enzyme</u>;
(Gene 2) allele B makes <u>enzyme</u> converting K to L / red to purple;
Allele b produces no / non-functional <u>enzyme</u>;
("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) F_1 are AaBb;
F_2 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] (a) Correct answer: 1.25;

21

Ignore working

OR (if wrong answer)

$$\frac{\text{measurement in µm}}{40000} \; / \; \frac{\text{measurement in mm}}{40} = 1 \text{ mark}$$

125 but wrong order of magnitude = 1 mark

2 (ii) **C** has myosin / thick (and actin / thin) filaments;

OR

A has only actin / thin (/ no myosin / no thick) filaments;

1 max

(b) When contracted:

Thick & thin filaments/myosin & actin overlap more;

Interaction between myosin heads & actin / cross-links form;

Movement of myosin head;

Thin filaments / actin moved along thick filaments / myosin;

Movement of thin filaments / actin pulls Z-lines closer together;

Displacement of tropomyosin to allow interaction;

Role of Ca^{2+};

Role of ATP;

Allow ref. to 'sliding filament mechanism' / described if no other marks awarded

4 max

(c) (i) 8 has DMD but 3 and 4 do not / 12 has DMD but 6 and 7 do not / neither parent has the condition but their child has;

Allow parents 3 and 4 give 8, parents 6 and 7 give 12

1

(ii) 4 ***AND*** 7;

1

(iii) Parental genotypes: 6 = $\mathbf{X^DY}$ AND 7 = $\mathbf{X^DX^d}$

AND

Gametes correct for candidate's P genotypes – e.g.

$\mathbf{X}^{D}$ and $\mathbf{Y} + \mathbf{X}^{D}$ and $\mathbf{X}^{d}$;

Offspring genotypes <u>correctly derived</u> from gametes e.g.

$\mathbf{X}^{D}\mathbf{X}^{D} + \mathbf{X}^{D}\mathbf{X}^{d} + \mathbf{X}^{D}\mathbf{Y} + \mathbf{X}^{d}\mathbf{Y}$;

Male offspring with MD correctly identified: $\mathbf{X}^{d}\mathbf{Y}$;

Probability = 0.25 / correct for candidates offsprings genotypes;

Accept ¼ / 1 in 4 / 1:3 / 25%

NOT '3:1' / '1:4'

4

(d) (i) No gene fragment **G**;

1

(ii) Only one copy of gene fragment **F**;

Male has only one X-chromosome / is XY
(c.f. female has two / is XX);

2

(iii) 10 has only one copy of gene fragment **G**;

10 has only one normal X-chromosome / has one abnormal / $X^{d}X^{D}X^{d}$ has only one normal allele / has one X / is X X / is heterozygous;

11 has two normal X-chromosomes / has 2 normal alleles / is $X^{D}X^{D}$ / has not got X^{d} / has 2 copies of (F and) G;

3

(e) (i) To prevent rejection / prevent antibody production vs. injected cells / injected cells have (foreign) antigen (on surface);

1

(ii) Shows effect of <u>cells</u> / not just effect of injection / not just effect of salt solution;

1

(iii) Only one person tested so far – need more to see if similar results /need more to see if reliable;

Need to assess if new (dystrophin positive) muscle fibres are functional / if muscle becomes functional;

Can't tell how widespread effect is in the muscle / sample taken near injection site;

Need to test for harmful side effects;

Need to test if successful for other mutations of dystrophin gene;

Need to assess permanence / longevity of result/insufficient time allowed in investigation;

(In this patient) only small response / %;

Further sensible suggestion;

4 max

[25] (a) Parents without CF → offspring with CF / 1 + 2 → 6 / 7 + 8 → I0;

22

Each parent must have CF allele / offspring receives CF allele from both parents / both parents heterozygous / both carriers;

2

(b) **Nn** and **Nn** (no mark since awarded in (a) already)

Accept alternative symbols

N n and **N n**;

Ignore X and Y

NN and **Nn** and **Nn** and **nn**;

Correct allocation of phenotypes to genotypes;

Probability = 0.125;

Accept answers expressed as chance rather than probability, eg 1 in 8 / 1 to 7 / 12.5%;

4

[6] (a) Daughter (C) does not have the condition / one child doesn't have it;

23

Accept converse arguments (If candidates see it purely as a genetic cross diagram) D is heterozygous because E is unaffected;

Parents must have been carriers of normal / healthy recessive/ if recessive then parents homozygous (so all children affected);

D has cancer, so the cancer allele must be dominant;

2

(b) Father (A) would pass on X chromosome to daughter;
She is not affected;

Accept that if D's X chromosome carried 'it', then E would be affected.

2

(c) Only 25 / young so don't know if cancer will develop;

Accept E must be homozygous recessive/have two recessive alleles;

Don't know if her father was heterozygous or homozygous;

So no chance of cancer / no more chance than rest of the population;

If heterozygous, she has a 50% chance of carrying the allele/gene;
If homozygous, she has a serious risk of cancer.

2 max

(d) Mutation / mutagen changes DNA of cell;
Damaged DNA not repaired / cells not killed / apoptosis doesn't happen; Mutation leads to loss of control / uncontrolled cell division;
(Some of these) cells carried to other parts of the body.

3 max

[9]