

GCSE Biology (9 – 1)

Family Trees and Genetic Diseases

Name:

Group:

Instructions

- Read each question carefully before you begin to answer it.
- Use black ink or black ball-point pen for your answers.
- Fill in the boxes at the top of this page with your name and group.
- Attempt to answer every question in the booklet.
- Answer the questions in the spaces provided – there may be more space than you need in some answer spaces.
- Calculators may be used for this booklet.
- For any calculations, you **must** show all your working.
- Any and all drawings or graphs should be completed in pencil.



For markers use	
Question	Mark
1	/6
2	/12
3	/12
4	/13
5	/11
6	/16
7	/9
Total	/79
%	
Grade	

Information

- The maximum mark for this paper is 79 marks.
- The marks for each question are shown in brackets.
- Please be aware that good English and clear presentation are required in your answers.



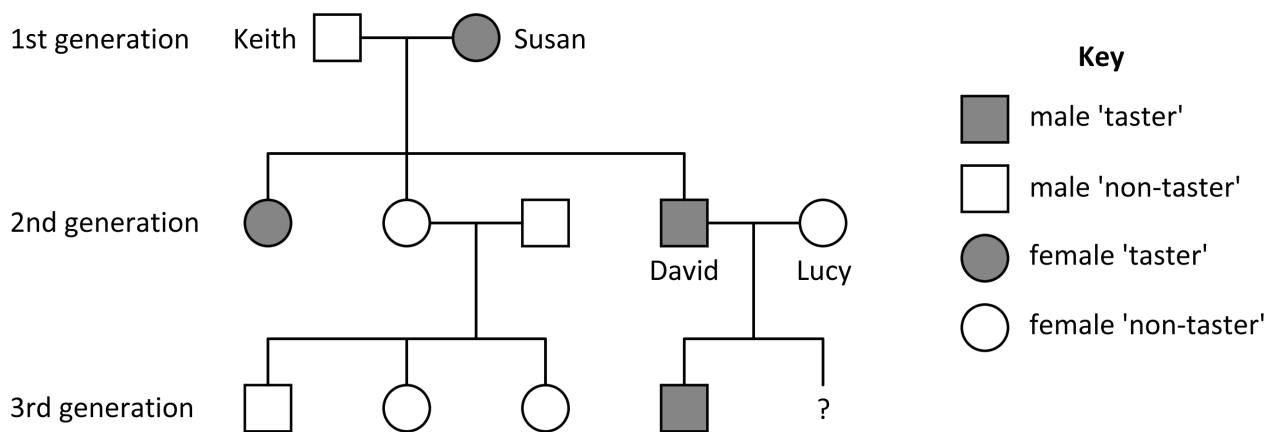
1. In biology, family trees or pedigrees serve as essential tools for tracing the inheritance patterns of genetic diseases through generations. Circle the letter of the correct answer for each.

a. What is always found in female gametes and may be found in male gametes?

- A one Y chromosome
- B one X chromosome
- C one X chromosome and one Y chromosome
- D two X chromosomes and two Y chromosomes

(1 mark)

b. This family tree shows how the ability to taste a certain substance is passed down. The allele enabling this taste is dominant, meaning it masks the effect of the allele for not being able to taste it.



What is the chance of the second child of David and Lucy being a 'non-taster'?

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|----------|----------|----------|----------|
| A | B | C | D |
| 1 in 1 | 1 in 2 | 1 in 3 | 1 in 4 |

(1 mark)

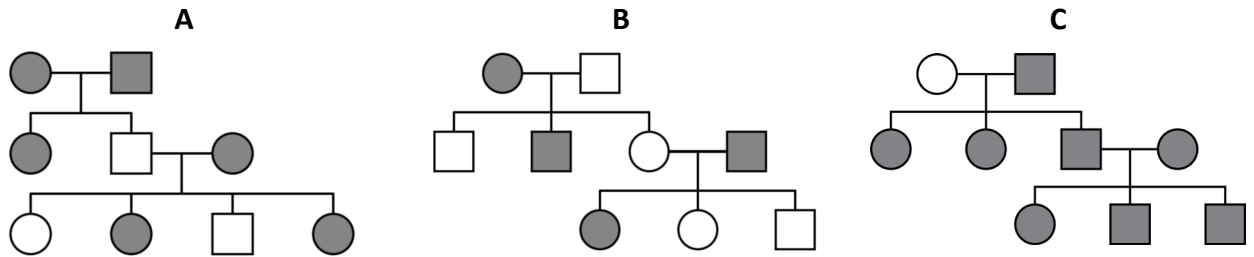
c. Cystic fibrosis is an inherited disorder caused by a recessive allele (f). The dominant allele (F) results in a healthy phenotype.

If two parents are **heterozygous** (Ff) for the cystic fibrosis allele, what is the probability that their first child will have cystic fibrosis?

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|----------|----------|----------|----------|
| A | B | C | D |
| 0% | 25% | 50% | 100% |

(1 mark)

- d. The family trees below show the inheritance of a **non sex linked** genetic disorder in three families. Which family tree best proves the disorder's dominant allele inheritance pattern.



(1 mark)

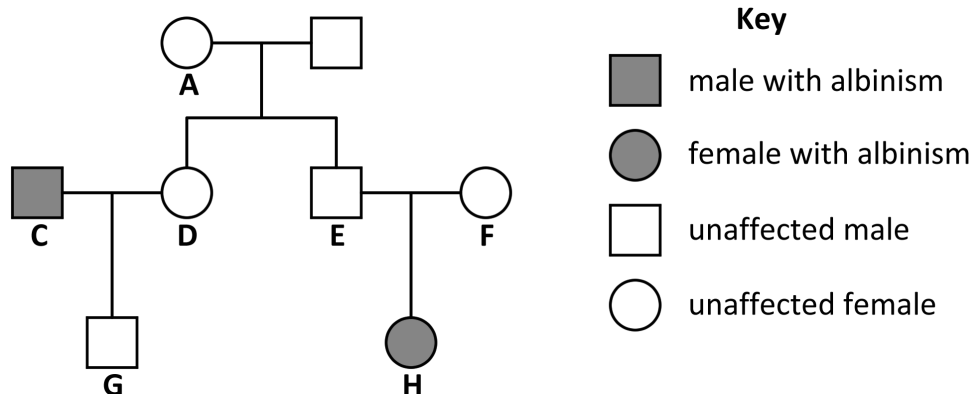
- e. Red-green colour blindness **is a sex-linked recessive condition**.
The allele for normal colour vision (R) is dominant to the allele for colour blindness (r).
These alleles are carried on the X chromosome.

A woman who is a carrier for colour blindness ($X^R X^r$) has a child with a man who has normal colour vision ($X^R Y$). What is the probability that their **son** will be red-green colour blind?

A	B	C	D
0%	25%	50%	100%

(1 mark)

- f. Albinism, an inherited condition caused by a recessive allele, is shown being passed down through one family in this pedigree diagram.



Which **labelled** individuals **must** be heterozygous for this condition?

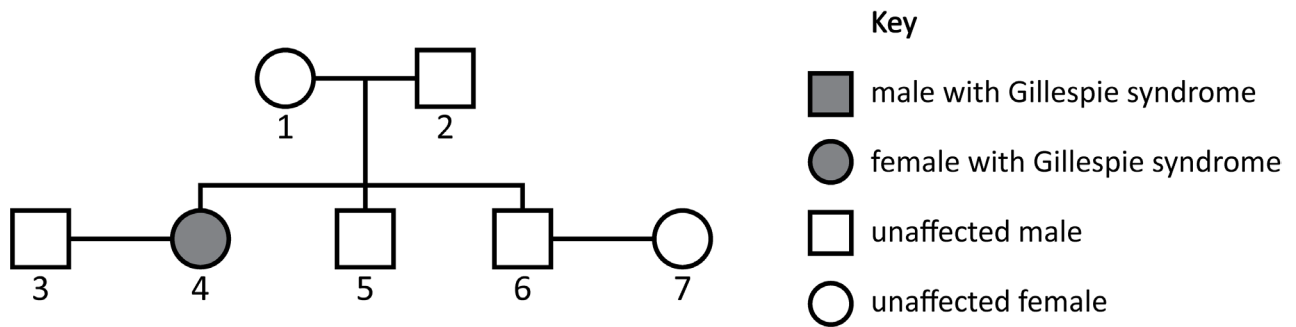
A	B	C	D
A and C	A, D and E	D, E and F	A, E, F and G

(1 mark)

(Total for Question 1 is 6 marks)

2. Gillespie syndrome is an uncommon hereditary disorder. Individuals with this condition typically present with eyes lacking an iris and experience damage to the cerebellum in the brain.

The mode of inheritance for Gillespie syndrome in a particular family is illustrated in the figure below.



- a. What does it mean if a genetic disorder is recessive?

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(2 marks)

- b. A genetic counselling makes this conclusion about Gillespie syndrome:

"The allele that causes Gillespie syndrome in this family is recessive".

Explain why this statement is correct. Use details from the figure above in your answer.

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(2 marks)

- c. Explain how the figure above shows that Person 1 and Person 2 are both heterozygous (carriers) for the Gillespie syndrome allele.

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(2 marks)

- d. Person 1 and person 2 are trying for another child.

Complete the Punnet square below and give the probability that the child will have Gillespie syndrome.

(Use **G** for the dominant allele and **g** for the recessive allele.)

Probability:

(3 marks)

- e. Persons 1 and 2 decide to have **two** more children after the one from the punnet square above.

What is the probability that both of these next two children will be **unaffected** by Gillespie syndrome?
Show your working.

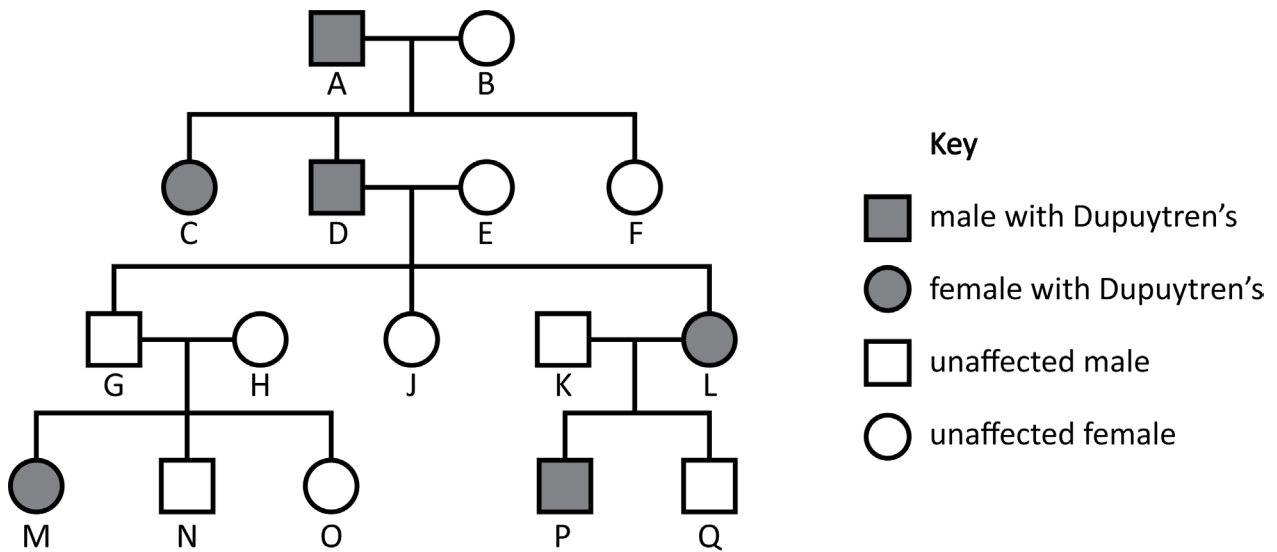
Probability = ...

(3 marks)

(Total for Question 2 is 12 marks)

3. Dupuytren's is a genetic disorder that primarily affecting the hands. The family tree diagram below shows how this condition is transmitted through one family.

Dupuytren's disorder is caused by a **dominant** allele.



- a. Explain how the figure above shows that Person A must be heterozygous for Dupuytren's. Use **D** to represent the dominant allele (causes Dupuytren's) and **d** to represent the recessive allele (normal). You should use a Punnett square in your answer.

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(3 marks)

- b. State the genotype of Person D.
Explain your answer using evidence from their parents (Person A and Person B).

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(2 marks)

- c. Person K and person L in the figure above are trying for a third child.
What is the probability of this third child having Dupuytren's when they are born?
In your answer you should:
- a Punnett square diagram
 - identify which offspring from the Punnett square have Dupuytren's.

Probability = ...
(5 marks)

- d. What evidence from the figure above demonstrates that the allele for Dupuytren's is **not** Y-linked

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(2 marks)

(Total for Question 3 is 12 marks)

4. Cystic fibrosis is an inherited disorder that affects cell membranes.
This leads to the production of unusually thick and sticky mucus by mucus-producing cells in many parts of the body, particularly affecting structures like the lungs, digestive system and reproductive system.

a) Explain how the production of thick, sticky mucus in individuals with cystic fibrosis can lead to difficulties with reproduction in **both** males and females.

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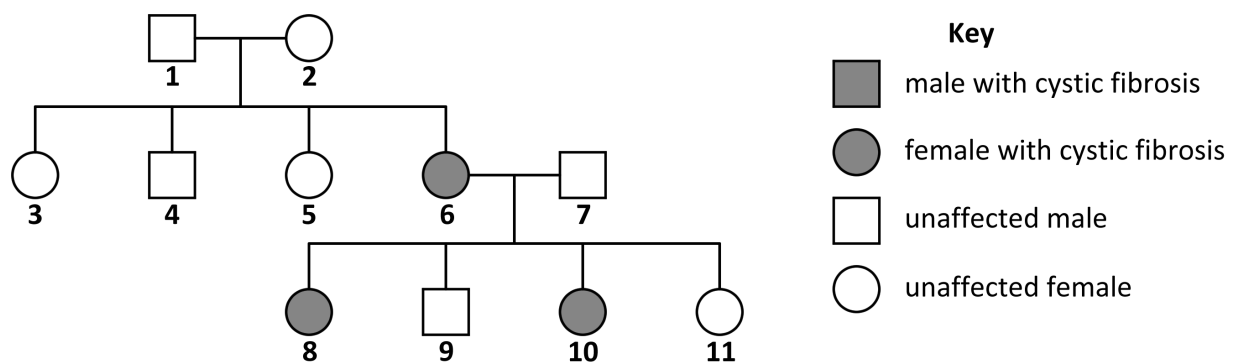
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(3 marks)

The family tree diagram below shows how cystic fibrosis is transmitted through one family.



b) Determine the genotype of person 7 and give a reason for your answer.

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(2 marks)

- c) Individuals 1 and 2 decide to have a fifth child.
Use a Punnet square to determine the probability that this child will **not** have cystic fibrosis.

Probability = ...
(4 marks)

- d) Explain why individuals with cystic fibrosis often experience:

- i. Frequent lung infections.

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(2 marks)

- ii. Difficulty digesting food and absorbing nutrients.

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(2 marks)

(Total for Question 4 is 13 marks)

5. Red-green colour blindness is a common genetic condition that affects how the eyes perceive colour. It is caused by a mutation to genes located on the X chromosome. This mutation results in an altered allele that is X linked recessive.

The condition affects specific cone cells in the retina, causing difficulty distinguishing between red and green colours.

a) Explain the meaning of these terms.

i. Allele:

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ii. Recessive:

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iii. X linked:

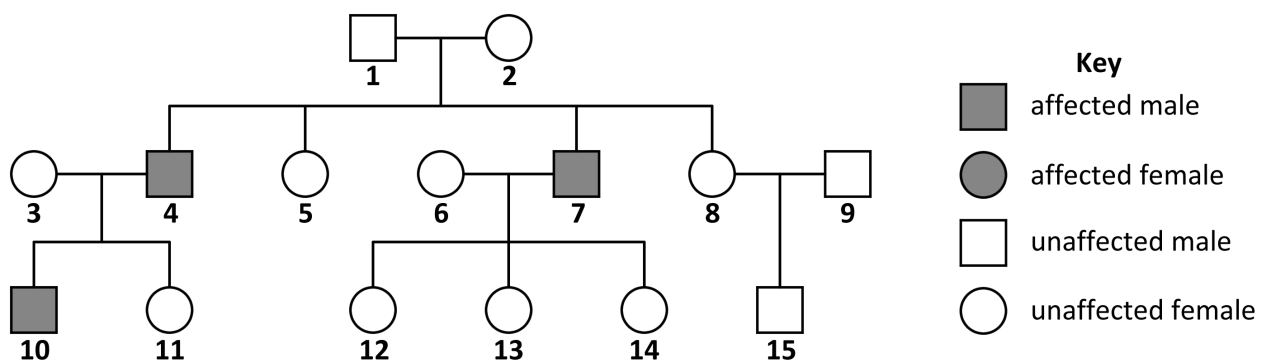
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(3 marks)

The family tree diagram below shows how red green colour blindness is transmitted through a family.



b) State the genotype of Person 2.

Explain your answer using evidence from their parents (Person 1 and Person 2).

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(2 marks)

- c) Individuals 6 and 7 are trying for another baby.

Draw a genetic diagram to calculate the probability that the baby will have the red green colour blindness.

Use X^R for the dominant allele and X^r for the recessive allele.

Probability = ...
(3 marks)

- d) Stem cells could potentially be used as used as a treatment for red green colour blindness in future.

- i. Explain why stem cells could be used as a treatment for red green colour blindness.

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 (2 marks)

- ii. Why is it an advantage to use stem cells from the individual with red green colour blindness rather than from another person?

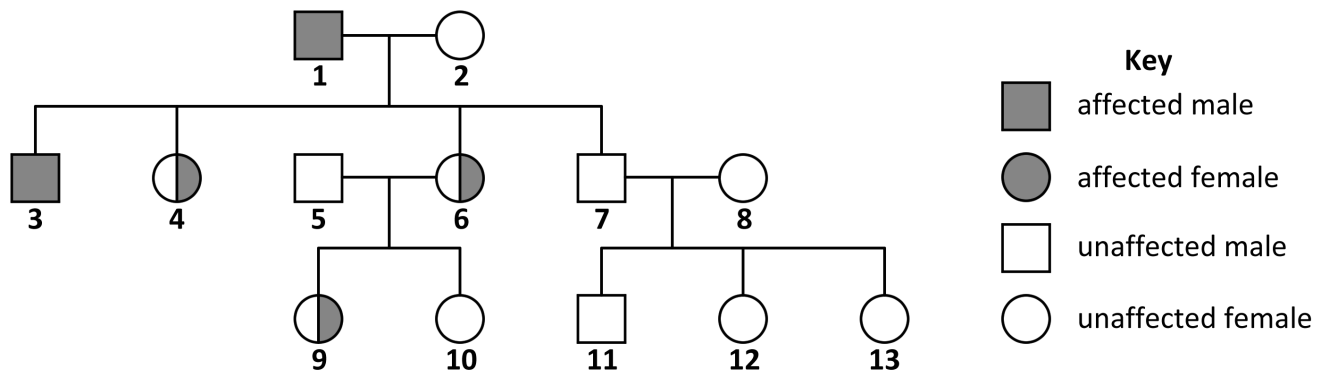
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 (1 mark)

(Total for Question 5 is 11 marks)

6. Haemophilia is a recessive sex linked disorder.

The family tree below shows the inheritance of haemophilia in a family.



a) What does the following symbol represent?



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(2 marks)

b) Explain why haemophilia is more common in males than females.

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(2 marks)

c) State the genotype of individual 1.

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(1 mark)

d) State the genotype of individual 2. Explain your answer.

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(2 marks)

e) Individuals 7 and 8 are trying for another child.

Explain, using details from the family tree diagram, the probability of individuals 7 and 8 having a child with haemophilia.

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(3 marks)

f) Individuals 5 and 6 are trying for another child.

Explain, using details from the family tree diagram, the probability of individuals 5 and 6 having a child with haemophilia.

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(3 marks)

g) State what haemophilia is, and describe how this condition can affect the human body.

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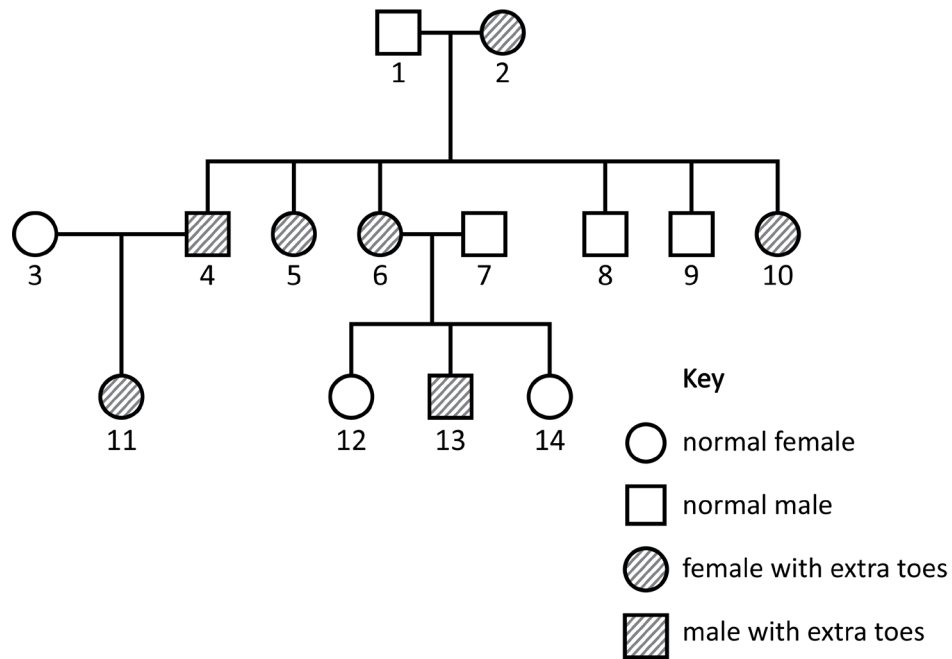
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(3 marks)

(Total for Question 6 is 16 marks)

7. A cat breeder has access to all the cats shown in the figure below.



- a) They want to start a breeding programme to produce a true-breeding line of cats that only have the normal number of toes. Using your knowledge of genetics, explain the plan the breeder should follow.

You must identify which cats from the pedigree they should use, and justify why their programme will eventually be successful in eliminating the allele for extra toes from their breeding line.

[illegible]

(6 marks)

b) The cross between cat 1 and cat 2 produced six offspring (cats 4, 5, 6, 8, 9 and 10).

If this same genetic cross were to produce a total of 20 kittens, calculate the number of kittens you would expect to have a normal number of toes. Show your working.

Expected number = ...

(2 marks)

c) The pedigree diagram shows that the actual number of unaffected offspring from the cross between cat 1 and cat 2 was 4.

Explain why the expected number of offspring from a genetic cross is often different from the actual number observed.

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(1 mark)

(Total for Question 7 is 9 marks)