

Family Trees and Genetic Diseases

Mark Scheme



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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?

B – is the only correct answer.	1 mark
	Total: 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'?

B – is the only correct answer.	1 mark
	Total: 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?

B – is the only correct answer.	1 mark
	Total: 1 mark

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

A – is the only correct answer.	1 mark
	Total: 1 mark

- e. Red-green colour blindness is a sex-linked recessive condition.

C – is the only correct answer.	1 mark
	Total: 1 mark

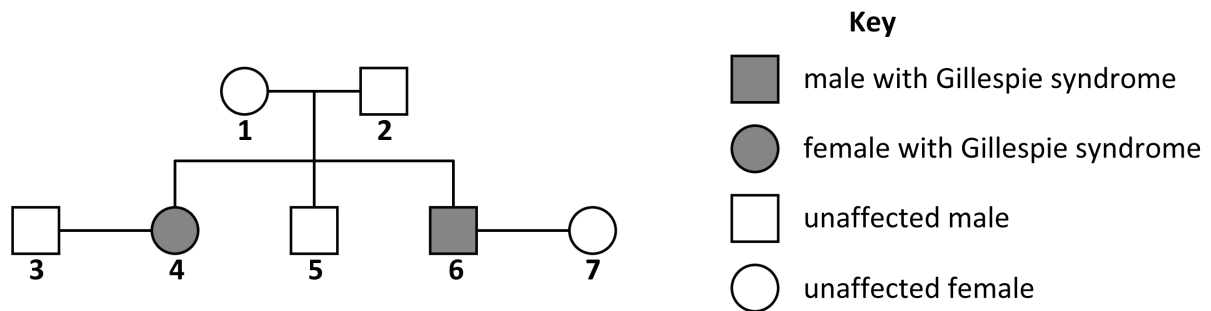
- f. Albinism, an inherited condition caused by a recessive allele, is shown being passed down through one family in this pedigree diagram. Which **labelled** individuals **must** be heterozygous for this condition?

D – is the only correct answer.	1 mark
	Total: 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 (aniridia) and experience damage to the cerebellum.

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?

An answer to include the following:	
Two copies of the allele are needed for the disorder to be expressed.	1 mark
The individual must be homozygous recessive (e.g., gg) to have the disorder.	1 mark
The allele is masked by / not expressed in the presence of a dominant allele.	1 mark
	Total: 2 marks

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

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

Individuals 1 and 2 are both unaffected.	1 mark
But they have produced affected offspring (4 and/or 6).	1 mark
	Total: 2 marks

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

An explanation to include the following:	
They are unaffected, so they must have at least one dominant / normal allele (G).	1 mark
They have affected children (4 and/or 6), so they must also carry the recessive / Gillespie syndrome allele (g) to pass on.	1 mark
	Total: 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.)

		G	g	
	G	GG	Gg	
	g	Gg	gg	
1 mark for the correct parent gametes.				
1 mark for the correct offspring genotypes.				
Award 1 mark for the correct probability: 0.25 or $\frac{1}{4}$ or 25%				
Total: 3 marks				

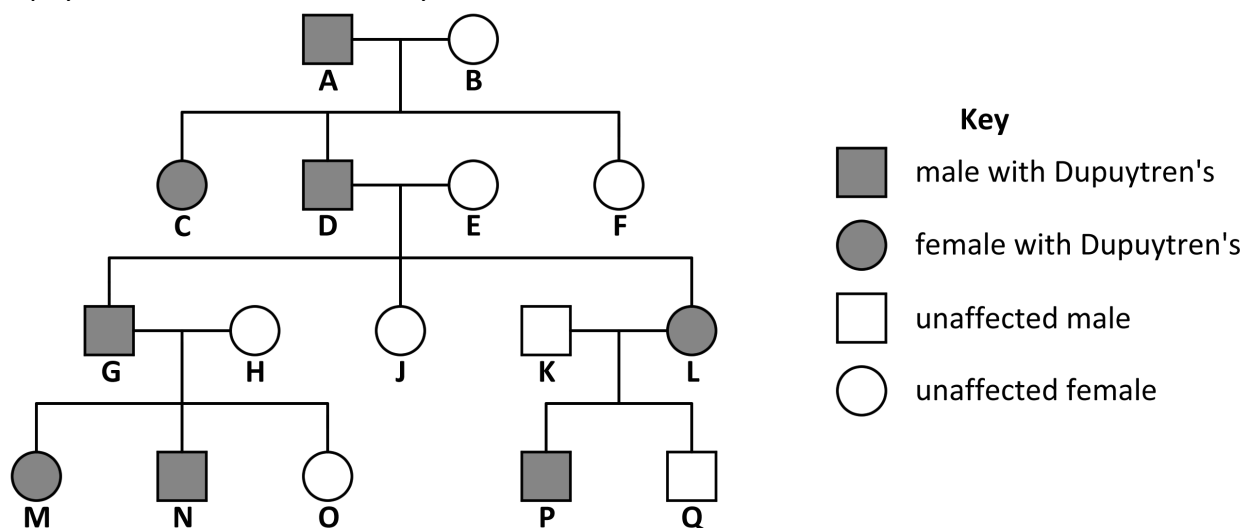
- e. Persons 1 and 2 decide to have **two** more children after the one from the punnet square above.
- What is the probability that both of these next two children will be **unaffected** by Gillespie syndrome?
Show your working.

Probability of one child being unaffected = $\frac{3}{4}$ or 0.75.	1 mark
Probability of two unaffected children = $(\frac{3}{4}) \times (\frac{3}{4})$ or 0.75×0.75 .	1 mark
Answer = $\frac{9}{16}$ (or 0.5625 or 56.25%)	1 mark
	Total: 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.

Person A and Person B have unaffected children (E and F) who must have the genotype dd.	1 mark
To have a dd child, they must inherit a recessive allele (d) from each parent, so Person A must carry the d allele.	1 mark
Since Person A is affected, they must have the dominant allele (D), therefore their genotype must be heterozygous (Dd).	1 mark
Marking note: A correct Punnett square for a Dd × dd cross, showing dd offspring are possible, can be awarded 1 mark if the explanation is weak.	
	Total: 3 marks

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

Genotype: Dd.	1 mark
Explanation: Person D inherited the dominant allele (D) from their affected parent (A) and a recessive allele (d) from their unaffected parent (B).	1 mark
	Total: 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.

		d	d	
	D	Dd	Dd	
	d	dd	dd	
Punnett square: Award 1 mark for the correct gametes of the parents. Award 2 marks for the correct genotypes of the offspring.				
Award 1 mark for the offspring with Dupuytren's identified: Dd				
Award 1 mark for the correct probability: $0.5 / \frac{1}{2} / 50\%$				
				Total: 5 marks

- d. What evidence in Figure 6 demonstrates that the allele for Dupuytren's is not Y-linked

Any one of the following points with explanation:	
Point: Affected male (A) has an unaffected son (E / F). Explanation: If the allele was Y-linked, an affected father would pass it to all his sons.	1 mark
Point: There are affected females (L / P). Explanation: Females do not have a Y chromosome, so cannot inherit a Y-linked disorder.	1 mark
	Total: 2 marks

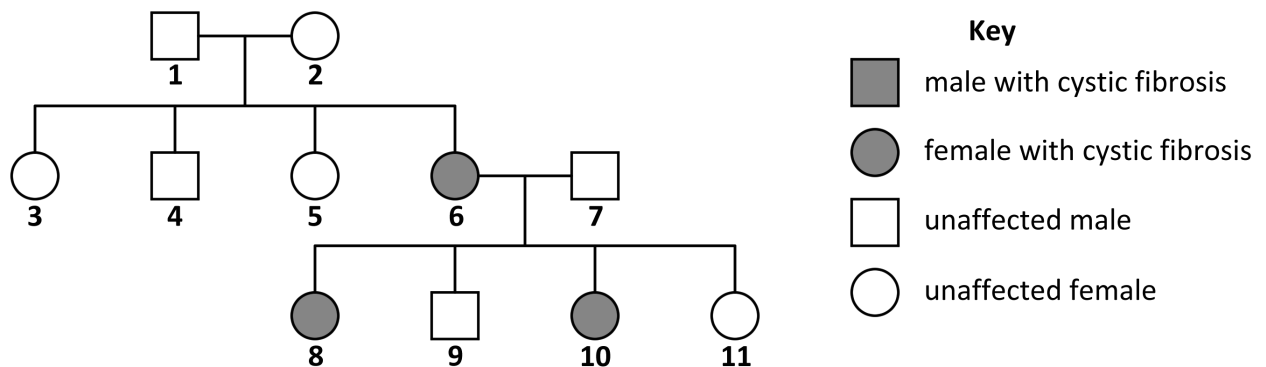
(Total for Question 3 is 12 marks)

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.

An explanation to include any three of the following:	
In males: Thick mucus blocks the sperm ducts / tubes. This prevents sperm from being released.	
In females: A thick mucus plug can form in the cervix. This prevents sperm from reaching the egg.	
	Total: 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.

Genotype: Ff (or heterozygous).	1 mark
Reason: Person 7 is unaffected, so has a dominant allele (F), but has affected children (8 and 10) so must also carry and have passed on the recessive allele (f).	1 mark
	Total: 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.

		F	f	
	F	Ff	Ff	
	f	Ff	ff	
Award 1 mark for the correct parent genotypes.				
Award 2 marks for the correct offspring genotypes.				
Award 1 mark for the correct probability: 0.75 / $\frac{3}{4}$ / 75%				
Total: 4 marks				

d) Explain why individuals with cystic fibrosis often experience:

i. Frequent lung infections.

Thick, sticky mucus in the airways is difficult for the cilia to move.	1 mark
This means that pathogens / bacteria are trapped in the lungs and can multiply, causing infection.	1 mark
	Total: 2 marks

ii. Difficulty digesting food and absorbing nutrients.

The tube from the pancreas (pancreatic duct) can become blocked by thick mucus.	1 mark
This prevents digestive enzymes from reaching the small intestine, so food is not properly broken down for absorption.	1 mark
	Total: 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:

An alternative / different form of a gene.	1 mark
	Total: 1 mark

ii. Recessive:

An allele that is only expressed (in the phenotype) if two copies are present / if there is no dominant allele.	1 mark
	Total: 1 mark

iii. X linked:

A gene / allele / trait that is located on the X chromosome.	1 mark
	Total: 1 mark

b) State the genotype of Person 2.

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

Genotype: $X^R X^r$	1 mark
Explanation: Person 2 is an unaffected female, but she has an affected son (Person 7). She must have passed the recessive allele (X^r) to him.	1 mark
	Total: 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.

		X^R	X^r		
	X^R	$X^R X^R$	$X^R X^r$		
	X^r	$X^R X^r$	$X^r X^r$		
Award 1 mark for the correct parent gametes.					1 mark
Award 1 mark for the correct offspring genotypes.					1 mark
Award 1 mark for the correct probability: $0.25 / \frac{1}{4} /$ or 25%					1 mark
					Total: 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.

An explanation to include the following:	
Stem cells are undifferentiated / unspecialised.	1 mark
They can be stimulated to differentiate into specialised cells, such as cone cells, to replace the faulty ones.	1 mark
	Total: 2 marks

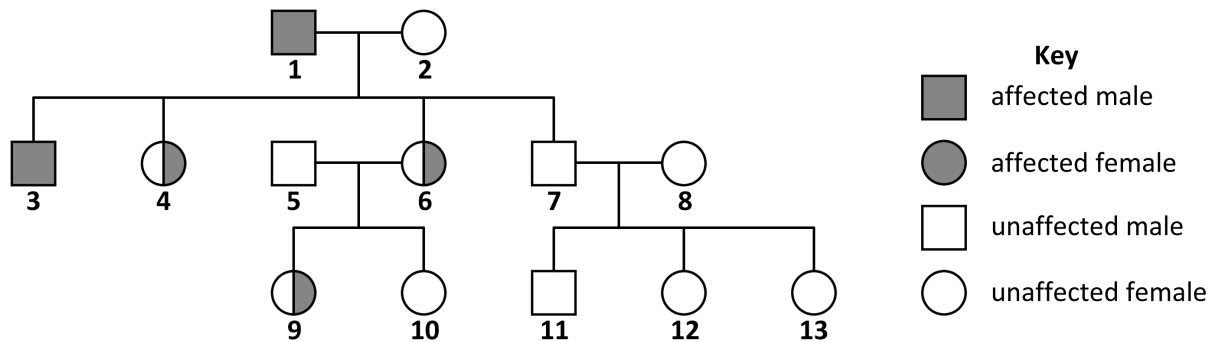
- ii. Why is it an advantage to use stem cells from the individual with red green colour blindness rather than from another person?

They are genetically identical to the patient's cells / have the same antigens, so they will not be rejected by the patient's immune system.	1 mark
Accept no need for immunosuppressant drugs.	
	Total: 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?

A female.	1 mark
Who is a <u>carrier of the condition</u> / they <u>carry the allele but are not affected by it</u> .	1 mark
	Total: 2 marks

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

The allele is on the X chromosome, and males only have one X chromosome.	1 mark
Therefore, males only need one copy of the recessive allele to be affected, whereas females need two copies.	1 mark
	Total: 2 marks

c) State the genotype of individual 1.

X^hY	1 mark
	Total: 1 mark

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

Genotype: X^HX^h	1 mark
Explanation: She is an unaffected female, but has an affected son (e.g., individual 3), so she must be a carrier for the recessive allele.	1 mark
	Total: 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.

An explanation to include any three of the following:	
Genotype of 7: X^HY (unaffected male). Genotype of 8: Can be X^HX^H or X^HX^h. - If 8 is a carrier (X^HX^h), the cross is $X^HY \times X^HX^h$, which gives a 1 in 4 (25%) chance of an affected child (X^hY). - If 8 is not a carrier (X^HX^H), the probability is 0.	
	Total: 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.

Genotype of 5: X^HY and Genotype of 6: X^HX^h .	1 mark
Punnett square showing the cross results in X^HX^h , X^hX^h , X^HY , X^hY .	1 mark
Probability: 0.25 / 1/4 / 25% (as two of the four outcomes, X^hX^h and X^hY , are affected).	1 mark
	Total: 3 marks

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

Haemophilia is a genetic disorder where the blood does not clot properly.	1 mark
This is due to a missing or faulty blood clotting factor / protein.	1 mark
This can cause prolonged bleeding after injury, internal bleeding, or painful joint damage.	1 mark
	Total: 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.

Marking guidance		
Level	Marks	Descriptor
Level 1	1 - 2	Simple, relevant statements are made, such as identifying that only cats with normal toes should be bred. The plan lacks detail, and the genetic explanation is limited or absent. The answer may not refer specifically to the pedigree chart.
Level 2	3 - 4	A viable plan is described, identifying the correct phenotype for breeding. Some suitable individuals from the pedigree may be named. The genetic reasoning is present but may lack detail or contain minor inaccuracies. The link between the breeding choices and the desired outcome is explained but not fully justified.
Level 3	5 - 6	A detailed and coherent plan is described. The genetic reasoning is clear, logical, and uses appropriate scientific terminology (homozygous recessive, alleles, true-breeding). Specific individuals from the pedigree are correctly identified as suitable for breeding.

Indicative Content:	
The Plan: - The breeder must select only cats with the normal number of toes for breeding. - Specific examples from the pedigree should be used, e.g., cat 1, cat 3, cat 7, cat 8, cat 9, cat 12, or cat 14. - These selected cats (e.g., cat 7 and cat 12) should be bred together. Genetic Justification: - The allele for extra toes is dominant. The allele for normal toes is recessive. - Therefore, any cat with the normal phenotype must have the genotype homozygous recessive (aa). - By selecting only normal-toed cats (aa), the breeder ensures that the parents do not possess the dominant allele for extra toes. - When two homozygous recessive (aa x aa) individuals are crossed, they can only pass on the recessive allele (a) to their offspring. - This means that all kittens produced will have the genotype aa and will therefore also have the normal number of toes. - This line of cats is now true-breeding for the normal-toed characteristic and the dominant allele has been eliminated.	
	Total: 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.

Identifies that the cross gives a probability of 0.5 (or 50% or 1 in 2) for normal toes. 20×0.5 or $20 \div 2$	1 mark
Answer: 10	1 mark
	Total: 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.

Fertilisation is a random process / it is down to chance.	1 mark
	Total: 1 mark

(Total for Question 7 is 9 marks)