AQA GCSE Biology | 8461

SPECIFICATION POINT 4.6.1.7

Inherited disorders

Past-paper questions

Tier	**Higher**
Question groups	**13**
Available paper years	**2008–2024**

Using this booklet

1. Answer the questions in order and show your working.
2. Original question numbers and marks are retained.
3. Print at actual size (100%) for measuring questions.
4. Use the companion answer booklet to check your work.

Name: _____ **Date:** _____

Whole questions are retained for context, so some parts assess other specification points. Current and legacy sources are labelled. Continue on extra paper if needed.

4.6.1.7 Inherited disorders

Question 01

June 2008 | BLY2H | Original Q2 | Legacy

2 Cystic fibrosis is an inherited disorder that can seriously affect health.

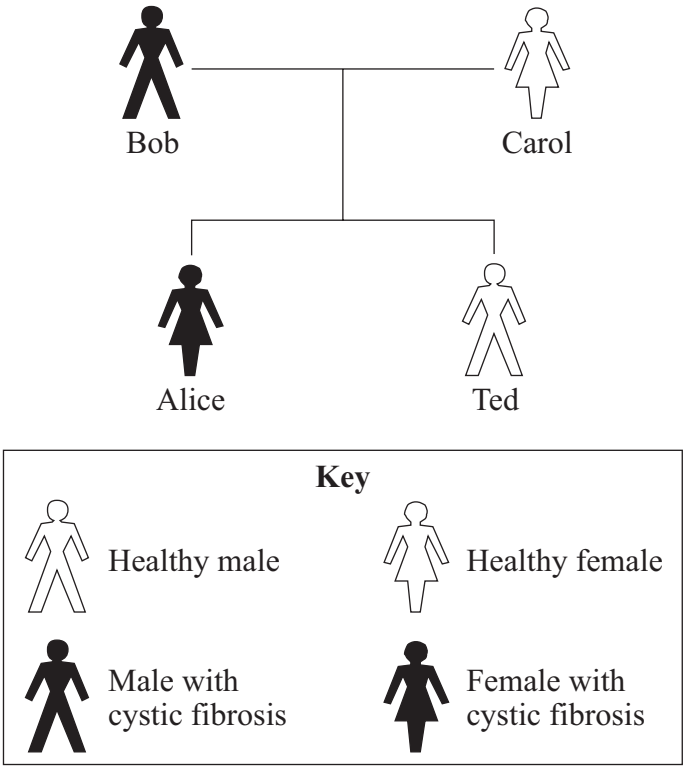
2 (a) Which **one** of these is affected by cystic fibrosis?

Draw a ring around your answer.

blood **cell membranes** **kidneys** **nervous system**

(1 mark)

2 (b) The diagram shows the inheritance of cystic fibrosis in a family. The allele that produces cystic fibrosis is recessive.



2 (b) (i) Explain why Alice inherited cystic fibrosis.

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(2 marks)

4.6.1.7 Inherited disorders

Question 01 · continued

for mark

2 (b) (ii) Explain why Ted did **not** inherit cystic fibrosis.

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(2 marks)

2 (c) Bob and Carol know that there is a risk that their next baby will have cystic fibrosis.

Embryos can be screened for the allele that produces cystic fibrosis.

Many people support the screening of embryos, but others do not.

2 (c) (i) Suggest **one** reason why many people support the screening of embryos for the cystic fibrosis allele.

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(1 mark)

2 (c) (ii) Suggest **one** reason why many people are against the screening of embryos for the cystic fibrosis allele.

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(1 mark)

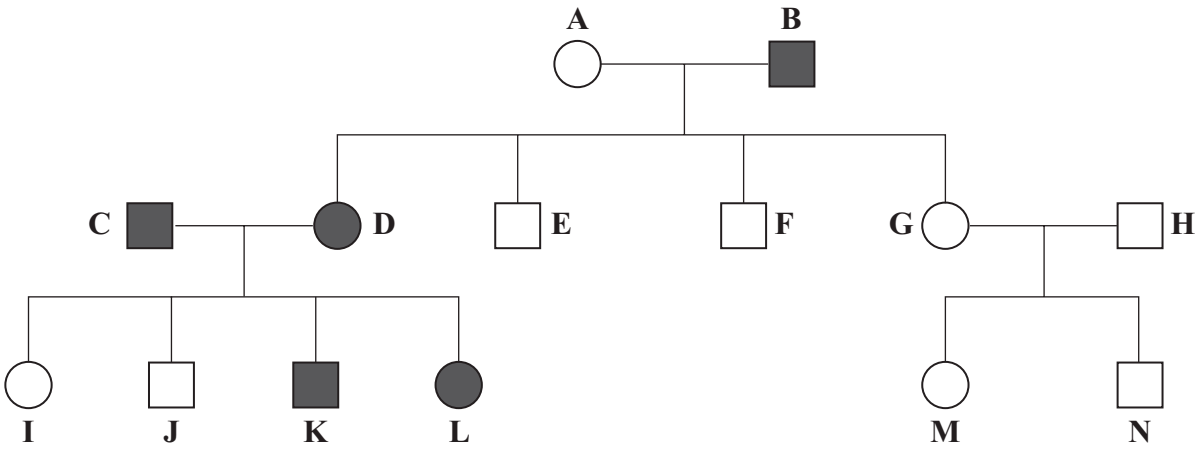
7

4.6.1.7 Inherited disorders

Question 02

January 2009 | BLY2H | Original Q6 | Legacy

6 The diagram shows a family tree in which some individuals have an inherited disorder, which may cause serious long-term health problems.



Key

Male without disorder

Male with disorder

Female without disorder

Female with disorder

6 (a) What proportion of the children of **A** and **B** have the disorder?

.....

(1 mark)

6 (b) Explain the evidence from the diagram which shows that the allele for the disorder is dominant.

Use the appropriate letters to identify individuals in your answer.

You may use genetic diagrams in your explanation. There is space for you to draw a genetic diagram at the top of the facing page.

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4.6.1.7 Inherited disorders

Question 02 · continued

for mark

(3 marks)

6 (c) (i) What is meant by 'embryo screening'?

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.....

(1 mark)

6 (c) (ii) A doctor suggests that couple **C** and **D** should have their embryos screened but that couple **G** and **H** do **not** need this procedure.

Explain the reasons for the doctor's suggestions.

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(3 marks)

8

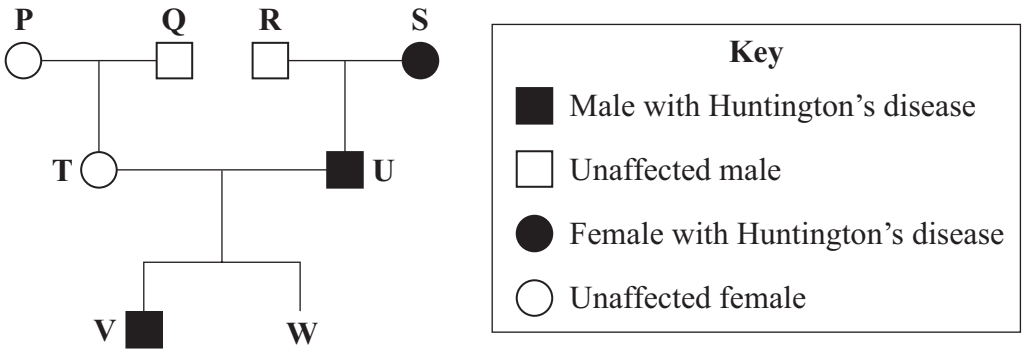
4.6.1.7 Inherited disorders

Question 03

June 2009 | BLY2H | Original Q6 | Legacy

6 Huntington's disease is a disorder of the nervous system. It is caused by a dominant allele.

The family tree shows the inheritance of Huntington's disease in one family.



6 (a) **T** and **U** already have a son, **V**, with Huntington's disease. What is the chance that their next child, **W**, will also develop Huntington's disease?

Use a genetic diagram in your answer.

Use the symbols **H** to represent the dominant allele and **h** to represent the recessive allele.

Chance that child **W** will have Huntington's disease = ...

(4 marks)

4.6.1.7 Inherited disorders

Question 03 · continued

for mark

6 (b) A doctor advises parents **T** and **U** that embryo **W** should be 'screened'.

6 (b) (i) What is embryo screening?

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.....

(1 mark)

6 (b) (ii) Some people do not believe that embryos should be screened.

Give **one** reason why.

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(1 mark)

6 (c) Cystic fibrosis is an inherited disorder in which the person's mucus becomes thick.

6 (c) (i) For this gene, one allele in every 30 in the population is the cystic fibrosis allele. However, only one person in every 900 has cystic fibrosis.

Explain why.

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(2 marks)

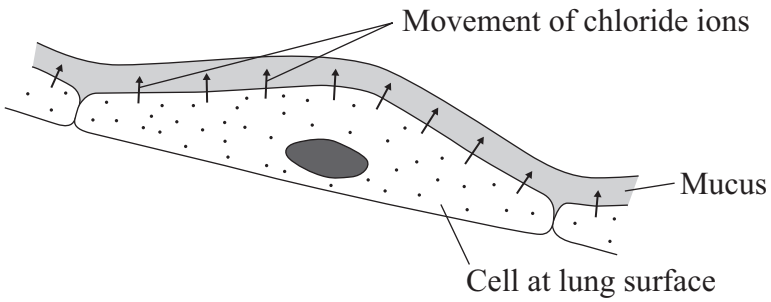
Question 6 continues on the next page

4.6.1.7 Inherited disorders

Question 03 · continued

for mark

6 (c) (ii) The diagram shows how, in a healthy person, cells at the lung surface move chloride ions into the mucus surrounding the air passages.



This movement of chloride ions causes water to pass out of the cells into the mucus.

Explain why.

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(3 marks)

6 (c) (iii) Mucus contains protein.

Which parts of the lung cells manufacture the protein in mucus?

.....

(1 mark)

12

4.6.1.7 Inherited disorders

Question 04

January 2010 | BLY2H | Original Q5 | Legacy

5 (a) Mr and Mrs Smith both have a history of cystic fibrosis in their families.
Neither of them has cystic fibrosis.
Mr and Mrs Smith are concerned that they may have a child with cystic fibrosis.

Use a genetic diagram to show how they could have a child with cystic fibrosis.

Use the symbol **A** for the dominant allele and the symbol **a** for the recessive allele.

(3 marks)

4.6.1.7 Inherited disorders

Question 04 · continued January 2010 | BLY2H | Original Q5 | Legacy

for mark

5 (b) Mr and Mrs Smith decided to visit a genetic counsellor who discusses embryo screening.

Read the information which they received from the counsellor.

- Under an anaesthetic five eggs will be removed from Mrs Smith's ovary.
- The eggs will be fertilised in a dish using Mr Smith's sperm cells.
- The embryos will be grown in the dish until each embryo has about thirty cells.
- One cell will be removed from each embryo and tested for cystic fibrosis.
- A suitable embryo will be placed into Mrs. Smith's uterus and she may become pregnant.
- Any unsuitable embryos will be killed.

5 (b) (i) Suggest why it is helpful to take five eggs from the ovary, rather than just one.

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(1 mark)

5 (b) (ii) Evaluate the use of embryo screening in this case.

Remember to give a conclusion as part of your evaluation.

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(4 marks)

8

4.6.1.7 Inherited disorders

Question 05

6 Cystic fibrosis and Huntington's disease are inherited disorders.

6 (a) Someone can be a carrier of cystic fibrosis.

Explain how.

You may include a genetic diagram in your answer.

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(2 marks)

6 (b) Why does only one parent need to have the Huntington's disease allele for a child to inherit Huntington's disease?

.....

.....

(1 mark)

3

4.6.1.7 Inherited disorders

Question 06

June 2012 | BL2HP | Original Q7 | Legacy

7 People with cystic fibrosis make large amounts of thick, sticky mucus in their lungs. Cystic fibrosis is caused by the inheritance of recessive alleles.

7 (a) What do each of the following mean?

7 (a) (i) Alleles

.....

.....

(1 mark)

7 (a) (ii) Recessive

.....

.....

(1 mark)

7 (b) Mr and Mrs Brown have a child with cystic fibrosis. They hope to have another child. They want to know the probability that their next child will have cystic fibrosis. They visit a genetic counsellor who explains, "You are both heterozygous for cystic fibrosis. There is a 1 in 4 (25%) chance that your next child will have cystic fibrosis."

Use the following symbols in answering the questions.

N = allele for being unaffected by cystic fibrosis
n = allele for cystic fibrosis

7 (b) (i) Mr and Mrs Brown both have the same genotype.

What is their genotype? .....

(1 mark)

4.6.1.7 Inherited disorders

Question 06 · continued

7 (b) (ii) There is a 1 in 4 chance that Mr and Mrs Brown's next child will have cystic fibrosis. Use a genetic diagram to explain why.

(3 marks)

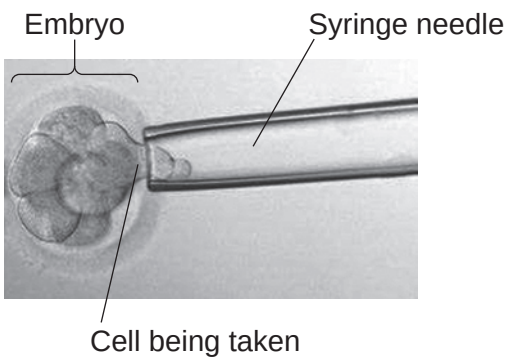
Question 7 continues on the next page

Question 06 · continued

7 (c) Mr and Mrs Brown do **not** want to have another child with cystic fibrosis. The genetic counsellor explains two different methods for finding out whether an embryo has cystic fibrosis. The methods are:

- pre-implantation genetic diagnosis (**PGD**)
- chorionic villus sampling (**CVS**).

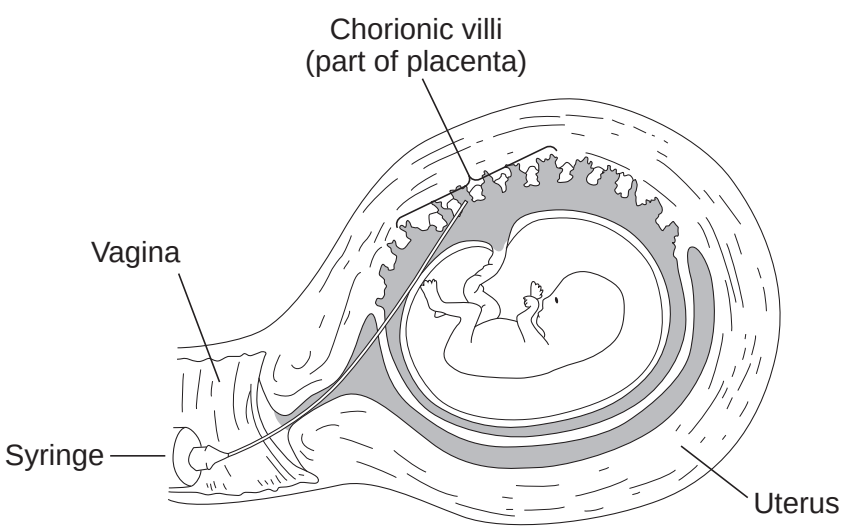
In **PGD**, eggs are fertilised in dishes and allowed to grow into embryos. A cell is taken from each embryo when the embryo is 3 days old. The photograph shows how the cell is taken.



Photograph: © Pascal Goetgheluck/Science Photo Library

The DNA in the cell can then be tested. The possibility of a false positive result is about 1 in 6. An unaffected embryo can then be placed in the woman's uterus. The procedure costs about £6000.

CVS can only be done after 9 weeks of pregnancy. A tiny piece of the placenta is taken out using a tube attached to a syringe. This is grown in tissue culture for about 7 days. The diagram below shows how **CVS** is done.



4.6.1.7 Inherited disorders

Question 06 · continued

The DNA in the cells can then be tested. About 2 in every 100 women have a miscarriage because of **CVS**. The possibility of a false positive result is about 1%. The procedure costs about £600. Following a positive result, the parents must then decide whether to terminate the pregnancy.

7 (c) The genetic counsellor thinks that **PGD** is a better method than **CVS** for detecting cystic fibrosis in an embryo.

Evaluate this opinion.

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(4 marks)

10

4.6.1.7 Inherited disorders

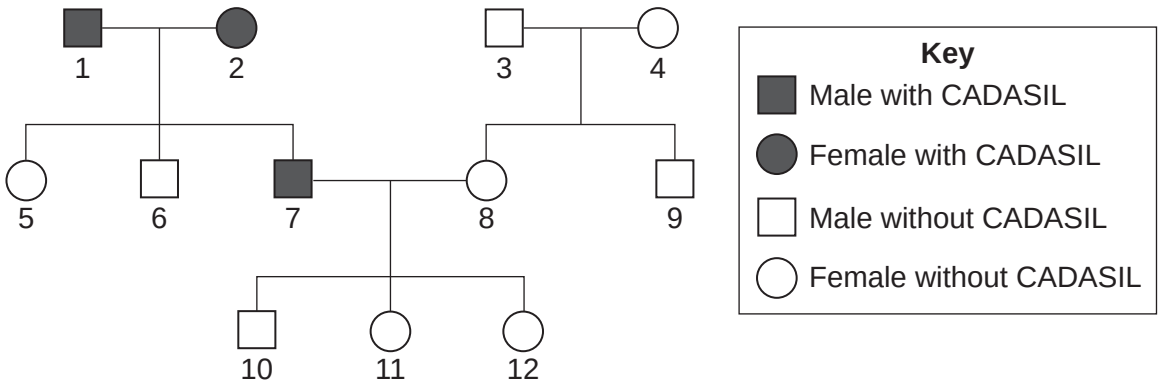
Question 07

January 2013 | BL2HP | Original Q6 | Legacy

6 CADASIL is an inherited disorder caused by a dominant allele.

CADASIL leads to weakening of blood vessels in the brain.

The diagram shows the inheritance of CADASIL in one family.



6 (a) CADASIL is caused by a *dominant allele*.

6 (a) (i) What is a *dominant allele*?

.....

.....

(1 mark)

6 (a) (ii) What is the evidence in the diagram that CADASIL is caused by a dominant allele?

.....

.....

(1 mark)

6 (a) (iii) Person **7** has CADASIL.

Is person **7** homozygous or heterozygous for the CADASIL allele?

Give evidence for your answer from the diagram.

.....

.....

(1 mark)

4.6.1.7 Inherited disorders

Question 07 · continued

6 (b) Persons **7** and **8** are planning to have another baby.
Use a genetic diagram to find the probability that the new baby will develop into a person with CADASIL.

Use the following symbols to represent alleles.

D = allele for CADASIL
d = allele for not having CADASIL

Probability = ...
(4 marks)

6 (c) Scientists are trying to develop a treatment for CADASIL using stem cells.

Specially treated stem cells would be injected into the damaged part of the brain.

6 (c) (i) Why do the scientists use stem cells?

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.....

.....

.....
(2 marks)

6 (c) (ii) Embryonic stem cells can be obtained by removing a few cells from a human embryo.

In 2006, scientists in Japan discovered how to change adult skin cells into stem cells.

Suggest **one** advantage of using stem cells from adult skin cells.

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(1 mark)

10

4.6.1.7 Inherited disorders

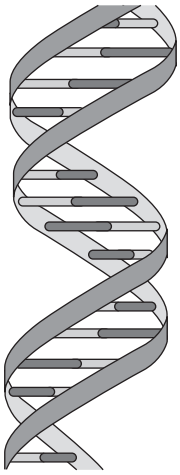
Question 08

June 2015 | BL2HP | Original Q3 | Legacy

3 DNA is the genetic material of human cells.

Figure 4 shows the structure of part of a DNA molecule.

Figure 4



3 (a) (i) Describe where DNA is found in a human cell.

[2 marks]

.....

.....

.....

3 (a) (ii) When a cell divides by mitosis the new cells are genetically identical.

What causes the cells to be genetically identical?

[1 mark]

.....

.....

3 (b) Many genes have different forms called alleles.

3 (b) (i) A person has polydactyly (extra fingers or toes). Polydactyly is caused by a dominant allele.
What is the smallest number of copies of the dominant allele for polydactyly that could be found in a body cell of this person?

[1 mark]

.....

3 (b) (ii) Another person has cystic fibrosis. Cystic fibrosis (CF) is caused by a recessive allele.
How many copies of the recessive CF allele are there in a body cell of this person?

[1 mark]

.....

4.6.1.7 Inherited disorders

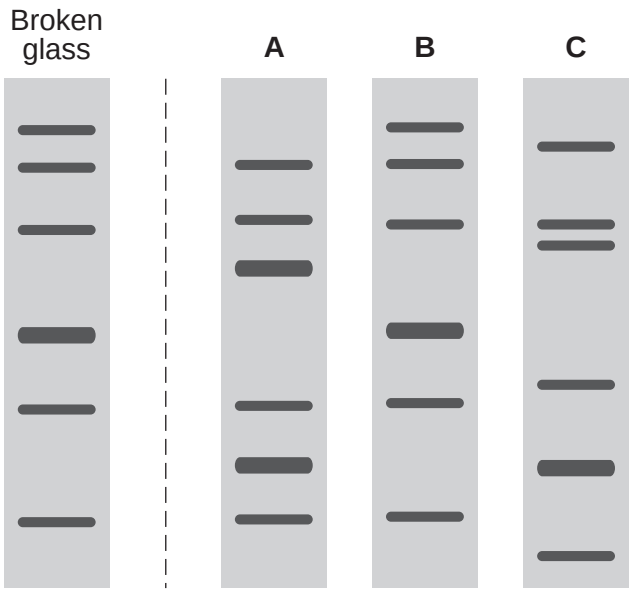
Question 08 · continued

3 (c) A burglar broke into a house. The burglar cut his hand on some broken glass. Scientists extracted DNA from the blood on the broken glass.

The scientists analysed the DNA from the glass and DNA from three suspects, **A**, **B** and **C**. The scientists used a method called DNA fingerprinting.

Figure 5 shows the scientists' results.

Figure 5



Which suspect, **A**, **B** or **C**, is most likely to have been the burglar?

[1 mark]

Tick (✓) **one** box.

A ☐

B ☐

C ☐

6

4.6.1.7 Inherited disorders

Question 09

June 2016 | BL2HP | Original Q6 | Legacy

6 (a) Polydactyly is an inherited condition caused by a dominant allele.

Figure 5 shows the hand of a man with polydactyly. The man has an extra finger on each hand.

The man's mother also has polydactyly but his father does not.

Figure 5



6 (a) (i) The man is **heterozygous** for polydactyly.

Explain how the information given above shows that the man is **heterozygous** for polydactyly.

[3 marks]

6 (a) (ii) The man marries a woman who does **not** have polydactyly.

What is the probability that their first child will have polydactyly?

[1 mark]

4.6.1.7 Inherited disorders

Question 09 · continued

6 (b) The man has red hair. His sister has brown hair.

Both of their parents have brown hair.

Brown hair is caused by the dominant allele, **B**.

Red hair is caused by a recessive allele, **b**.

Complete the genetic diagram below to show how the man's parents were able to have some children with red hair and some with brown hair.

[5 marks]

	Father	**Mother**
Parental phenotypes:	_____	_____
Parental genotypes:	_____	_____
Gametes:	_____ _____	_____ _____

Offspring genotypes: _____

Offspring phenotypes: _____

9

4.6.1.7 Inherited disorders

Question 10

June 2018 | 8461/1H | Original Q6

0 6 Cystic fibrosis (CF) is a genetic disorder caused by a change in a gene.

0 6 . 1 What molecule are genes made of?

[1 mark]

0 6 . 2 CF affects the cell membranes of cells in the lungs and digestive system.

What is the function of the cell membrane?

[1 mark]

0 6 . 3 In a person with CF, cells lining the lungs and digestive system create too much mucus.

The mucus can:

- block the duct leading from the pancreas to the small intestine
- block the tubes leading to the alveoli in the lungs.

Explain why children with CF grow more slowly than children without CF.

[6 marks]

4.6.1.7 Inherited disorders

Question 10 · continued

Table 6 shows information about people in the UK in 2015.

Table 6

	Median age in years
People with CF	19
Whole population	40

0 6 . 4 Describe how the median age of a group of people can be determined.

[2 marks]

0 6 . 5 Suggest **one** reason why the median age for people with CF is lower than the median age for the whole population.

[1 mark]

Question 10 · continued

0 6 . 6 People with a lung function below 30% may need a lung transplant.

Table 7 gives information about people with CF in 2015.

Table 7

Lung Function (%)	Percentage of people with CF
>75	22
51 – 75	72
30 – 50	4
<30	2

In 2015, the total number of people with CF in the UK was 10 800.

Calculate how many people with CF in the UK in 2015 would **not** need a lung transplant.

[2 marks]

Number of people = _____

4.6.1.7 Inherited disorders

Question 10 · continued

0 6 . 7 Lung transplants from donors have risks. One risk is organ rejection.

Scientists are researching how to solve the problem of organ rejection and hope to use stem cells to create healthy lungs.

The healthy lungs can then be transplanted into CF patients without the risk of organ rejection.

Describe how scientists may use stem cells to create healthy lungs that are **not** rejected by the CF patient.

[4 marks]

0 6 . 8 Some people disagree with the use of stem cells because of the risk of cancer.

Give **one** other reason why some people disagree with the use of stem cells to create new organs for transplants.

[1 mark]

18

4.6.1.7 Inherited disorders

Question 11

November 2021 | 8461/2H | Original Q8

0 8 Sickle cell anaemia is an inherited condition that affects red blood cells.

Sickle cell anaemia is caused by a mutation in the gene for haemoglobin. Haemoglobin is the red pigment found in red blood cells.

A person who is homozygous for the normal haemoglobin allele (H^A) produces normal red blood cells.

A person who is homozygous for the mutated allele (H^S):

- produces red blood cells with abnormal haemoglobin
- has red blood cells that can form an altered shape
- has sickle cell anaemia and becomes ill.

A person who is heterozygous:

- has both normal and abnormal haemoglobin in the red blood cells
- has sickle cell trait
- is generally healthy but can become ill in certain circumstances.

0 8 . 1 Give the reason why a mutation in the gene coding for haemoglobin could be harmful.

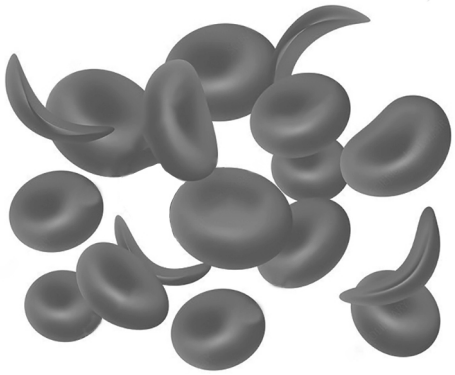
[1 mark]

4.6.1.7 Inherited disorders

Question 11 · continued

0 8 . 2 **Figure 13** shows some red blood cells from the blood of a person with sickle cell trait.

Figure 13



Calculate the proportion of cells in **Figure 13** that have an altered shape.

[2 marks]

Proportion = _____

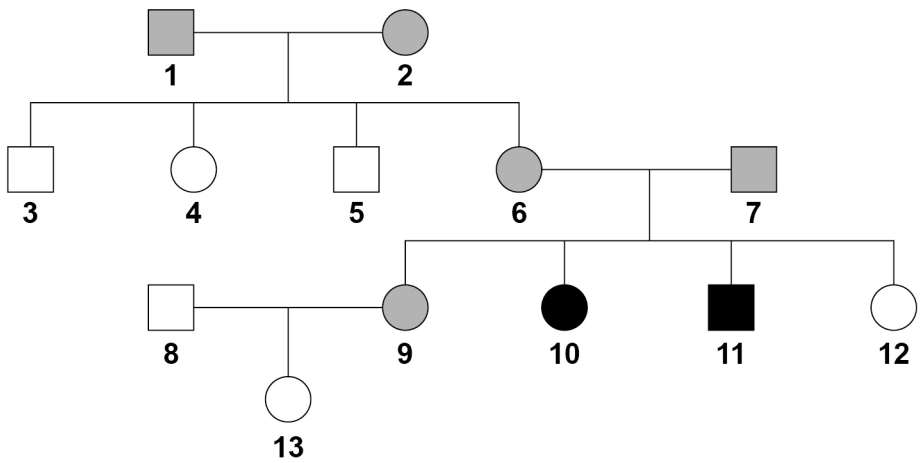
Question 8 continues on the next page

4.6.1.7 Inherited disorders

Question 11 · continued

Figure 14 shows the inheritance of sickle cell anaemia in one family.

Figure 14



Key

- Unaffected male
- Unaffected female
- Male with sickle cell anaemia
- Female with sickle cell anaemia
- Male with sickle cell trait
- Female with sickle cell trait

4.6.1.7 Inherited disorders

Question 11 · continued

0 8 . 3 Persons **8** and **9** in **Figure 14** are expecting a second child.

Determine the probability that the child will be a girl with sickle cell trait.

You should:

- draw a Punnett square diagram
- identify the phenotype of each offspring genotype
- use the symbols:

  H^A = normal haemoglobin allele

  H^S = mutated haemoglobin allele.

[5 marks]

Probability of a girl with sickle cell trait = _____

Question 8 continues on the next page

4.6.1.7 Inherited disorders

Question 11 · continued

0 8 . 4 Without medical treatment, people with sickle cell anaemia are frequently ill and have a reduced life expectancy.

The malarial parasite cannot live in the red blood cells of a person who has the $\mathbf{H^S}$ allele.

A scientist stated:

'It is an advantage for people to have the $\mathbf{H^S}$ allele in countries where malaria occurs.'

Evaluate the scientist's statement.

[3 marks]

11

4.6.1.7 Inherited disorders

Question 12

June 2024 | 8461/1H | Original Q2

0 2 Cystic fibrosis (CF) is an inherited disorder caused by a faulty gene.

0 2 . 1 Where in a cell would the CF gene be found?

[1 mark]

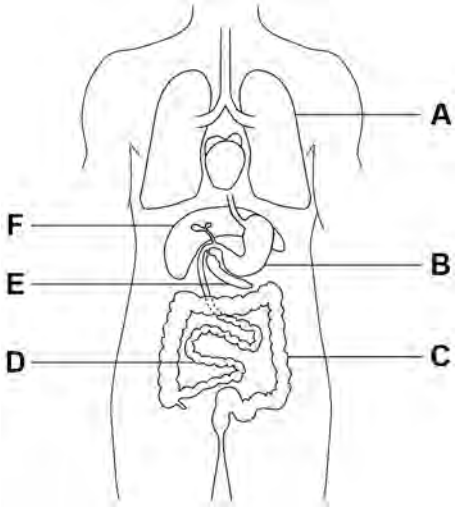
CF affects many organs in the body.

The main organs affected are:

- the lungs
- the pancreas
- the small intestine.

0 2 . 2 **Figure 2** shows organs of the human body.

Figure 2



Which letters in **Figure 2** show the lungs, the pancreas and the small intestine?

[1 mark]

Tick (✓) **one** box.

A, **D** and **E** ☐

A, **E** and **F** ☐

B, **C** and **D** ☐

B, **C** and **F** ☐

Question 2 continues on the next page

4.6.1.7 Inherited disorders

Question 12 · continued

0 2 . 3 The pancreas produces several digestive enzymes.

CF reduces the amount of each enzyme that reaches the small intestine.

Explain why a person with CF has:

- difficulty digesting food
- difficulty gaining body mass.

[6 marks]

4.6.1.7 Inherited disorders

Question 12 · continued

0 2 . 4 Gas exchange happens in the alveoli in the lungs.

Describe **three** features of the alveoli that help maximise gas exchange.

[3 marks]

1 _____

_____

2 _____

_____

3 _____

_____

0 2 . 5 CF reduces the amount of oxygen that can enter the blood from the alveoli.

Explain how a reduced amount of oxygen entering the blood will affect the human body.

[3 marks]

_____

_____

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_____

14

4.6.1.7 Inherited disorders

Question 13

0 6 Some human disorders are inherited.

Polydactyly is an inherited disorder.

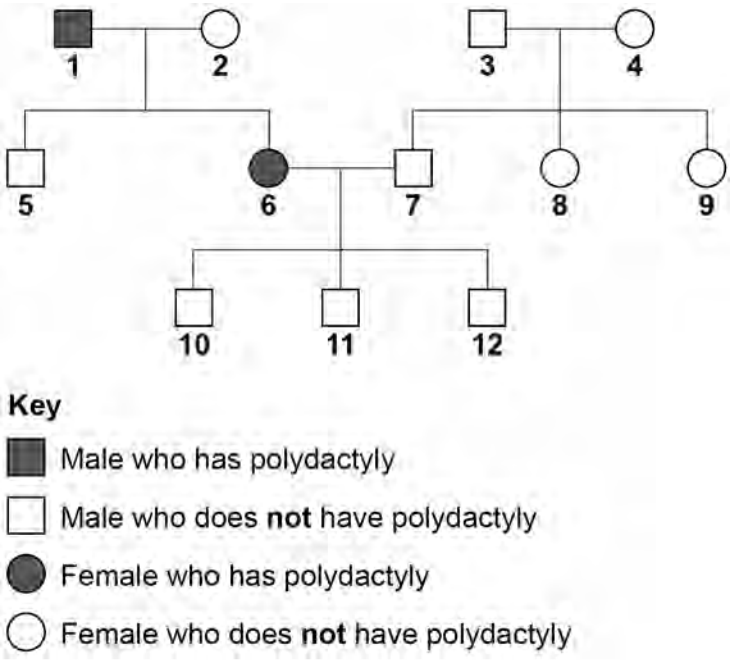
- A person with polydactyly has extra fingers or toes.
- Polydactyly is caused by a dominant allele.

0 6 . 1 What is a dominant allele?

[1 mark]

Figure 10 shows the inheritance of polydactyly in one family.

Figure 10



4.6.1.7 Inherited disorders

Question 13 · continued

In questions **06.2** and **06.3**, use the following symbols:

D = allele for having polydactyly

d = allele for **not** having polydactyly.

0 6 . 2 Person **1** is heterozygous.

Explain how **Figure 10** shows that person **1** is heterozygous.

[2 marks]

0 6 . 3 Persons **6** and **7** are expecting a fourth child.

A doctor states that the probability of having a child with polydactyly is 0.5

Explain how the doctor determined this probability.

[4 marks]

You should:

- draw a Punnett square diagram
- give the genotype of person **6** and the genotype of person **7**
- identify **all** the offspring that will have polydactyly.

Question 6 continues on the next page

4.6.1.7 Inherited disorders

Question 13 · continued

0 6 . 4 Cystic fibrosis (CF) is another inherited disorder caused by a mutation.

The mutation occurs in a gene called CFTR.

For the CFTR gene, one **allele** in every 50 in the UK population is the cystic fibrosis allele.

Explain why only one person in 2500 in the UK population has cystic fibrosis.

[4 marks]

11