

GCSE Combined Science: Trilogy

Worksheet: Inherited disorders

TIME 30 min	TOTAL 30 marks	CALCULATOR Allowed	DIFFICULTY Easy, Medium
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Topics: Inherited disorders

NAME	CLASS	DATE
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- Use black ink or black ball-point pen. Pencil should only be used for drawing.
- Answer all questions in the spaces provided.
- In all calculations, show clearly how you work out your answer.
- The marks for questions are shown in brackets.

0 1

A couple are both carriers of cystic fibrosis. They are thinking about using IVF with embryo screening.

0 1 . 1 Which statement about a carrier of cystic fibrosis is correct?
Tick (✓) **one** box.

A carrier cannot pass the allele on to their children

☐

A carrier has cystic fibrosis

☐

A carrier has one copy of the recessive allele and does not have the disorder

☐

A carrier has two copies of the recessive allele

☐

[1 mark]

0 1 . 2 Give **one** argument for embryo screening.

.....

.....

[1 mark]

0 1 . 3 Give **one** argument against embryo screening.

.....

.....

[1 mark]

0 1 . 4

Polydactyly is caused by a dominant allele.

How many copies of this allele must a person inherit to have polydactyly?

.....

.....

.....

[1 mark]

0 2

Some disorders are caused by the inheritance of recessive alleles.

0 2 . 1

Explain why two parents who do not have a genetic disorder can have a child who does.

.....

.....

.....

.....

[2 marks]

0 2 . 2

A recessive allele that causes a disorder can seem to 'skip' a generation.

Which statement explains this?

Tick (✓) **one** box.

Carriers show mild symptoms of the disorder

☐

The allele mutates back and forth between generations

☐

The allele can be passed on by unaffected carriers for generations

☐

Recessive alleles are chemically unstable

☐

[1 mark]

0 2 . 3

Cystic fibrosis is caused by a recessive allele.
Give the genotype of a person who has cystic fibrosis. Use F for the dominant allele and f for the recessive allele.

.....

.....

.....

Genotype =

[1 mark]

0 3

Some disorders are inherited. They are caused by the inheritance of certain alleles.

0 3 . 1

What causes polydactyly?

Tick (✓) **one** box.

A bacterial infection

☐

A dominant allele

☐

A poor diet

☐

A recessive allele

☐

[1 mark]

0 3 . 2

Describe what polydactyly is.

.....

.....

[1 mark]

0 3 . 3

Cystic fibrosis is a disorder of which part of the cell?

Tick (✓) **one** box.

Cell membrane

☐

Cell wall

☐

Mitochondria

☐

Ribosomes

☐

[1 mark]

0 3 . 4

Cystic fibrosis is caused by a recessive allele.
What word describes a person who has one copy of this allele but does not have the disorder?

.....

.....

.....

.....

[1 mark]

0 3 . 5

Human embryos can be tested for the alleles that cause genetic disorders.
What is this process called?

.....

.....

[1 mark]

0 4

Cystic fibrosis (CF) is caused by a recessive allele, f. The dominant allele, F, results in normal cell membranes. Two parents are both carriers of CF.

0 4 . 1

Give the genotype of a carrier of CF.

.....

.....

[1 mark]

0 4 . 2

Table 1

Gametes	F	f
F		
f		

Complete the Punnett square in Table 1 to show the possible genotypes of the couple's children.

[2 marks]

0 4 . 3

What is the probability that a child of this couple will have CF?

.....

.....

[1 mark]

0 4 . 4

What is the probability that a child of this couple will be a carrier of CF?

.....

.....

[1 mark]

0 4 . 5

The couple's first child has CF.
Explain why the probability that their second child will have CF is still the same.

.....

.....

[1 mark]

0 5

In a certain population, 1 in 25 people are carriers of a recessive allele that causes an inherited disorder.

0 5 . 1

Calculate the percentage of the population who are carriers.

.....

.....

.....

Percentage = %

[1 mark]

0 5 . 2

Out of 12 000 people in a town, estimate how many would be expected to be carriers.

.....

.....

.....

.....

Number of carriers =

[2 marks]

0 5 . 3

Explain why being a carrier does not affect a person’s own health.

.....

.....

.....

.....

[2 marks]

.....

0 6

A health programme offered free carrier testing for a recessive disorder to two groups: people with a family history of the disorder, and people with no known family history. Table 2 shows the results.

Table 2

Group	Number offered testing	Number who had the test	Number found to be carriers
family history	400	340	68
no family history	5000	850	34

0 6 . 1

Calculate the percentage of each group who chose to have the test.
Give your answers in the order: family history, no family history.

.....

.....

.....

.....

.....
[2 marks]

0 6 . 2

Calculate the percentage of the people tested who were found to be carriers, in each group.
Give your answers in the order: family history, no family history.

.....

.....

.....

.....

.....
[2 marks]

0 6 . 3

Suggest why people with a family history of the disorder were both more likely to accept testing, and more likely to be found to be carriers.

.....

.....

.....

.....

[2 marks]

END OF QUESTIONS

Original practice questions written to match the specification. Not produced by, affiliated with or endorsed by AQA.

Mark scheme

GCSE Combined Science: Trilogy · Worksheet: Inherited disorders · 30 marks

Question 1 (4 marks)

01.1 [1 mark]

Correct: A carrier has one copy of the recessive allele and does not have the disorder

01.2 [1 mark]

Answer: It prevents a child being born with a serious disorder, avoiding suffering

- prevents suffering / a child being born with the disorder / reduces the cost of care / lets carriers have a child without the disorder (1)
-

01.3 [1 mark]

Answer: Embryos with the disorder are destroyed, and some people believe embryos have a right to life

- embryos are destroyed / embryos have a right to life / expensive / could lead to 'designer babies' / the test may not be accurate (1)
-

01.4 [1 mark]

Answer: One

- one / 1 (1)

Notes: Allow 'one or two' / 'at least one'.

Question 2 (4 marks)

02.1 [2 marks]

Answer: Both parents can be (unaffected) carriers of the recessive allele, and each can pass it on to the child, who then has two copies of the allele

- both parents can be (unaffected) carriers of the recessive allele (1)
- each parent can pass it on to the child, who then has two copies (is homozygous recessive) (1)

02.2 [1 mark]

Correct: The allele can be passed on by unaffected carriers for generations

02.3 [1 mark]

Answer: ff

- ff (1)

Question 3 (5 marks)

03.1 [1 mark]

Correct: A dominant allele

03.2 [1 mark]

Answer: Having extra fingers or toes

- having extra fingers or toes (1)

03.3 [1 mark]

Correct: Cell membrane

03.4 [1 mark]

Answer: Carrier

- carrier (1)

03.5 [1 mark]

Answer: Embryo screening

- embryo screening (1)

Notes: Allow genetic screening / genetic testing of embryos.

Question 4 (6 marks)

04.1 [1 mark]

Answer: Ff

- Ff (1)

04.2 [2 marks]

Answer: FF, Ff, Ff, ff

- FF and ff in the correct boxes (1)
- Ff in both of the other boxes (1)

04.3 [1 mark]

Answer: 0.25 (25%, $\frac{1}{4}$)

- 0.25 / 25% / $\frac{1}{4}$ / 1 in 4 (1)

Notes: Do not accept 1 : 4 or 1 : 3.

04.4 [1 mark]

Answer: 0.5 (50%, $\frac{1}{2}$)

- 0.5 / 50% / $\frac{1}{2}$ / 1 in 2 (1)

04.5 [1 mark]

Answer: Each fertilisation is a separate random event, so the result for one child does not affect the next

- each fertilisation is independent / random, so the probability is the same for every child (1)
-

Question 5 (5 marks)

05.1 [1 mark]

Answer: 4%

- $1 \div 25 \times 100 = 4 \text{ (\%)} \text{ (1)}$

05.2 [2 marks]

Answer: 480

- $12\,000 \div 25 \text{ (1)}$
- 480 (1)

Notes: Correct answer with no working scores 2 marks.

05.3 [2 marks]

Answer: A carrier has one recessive allele and one dominant allele; the dominant allele is expressed, so the recessive allele's effect is masked

- a carrier has one recessive allele and one dominant allele (1)
 - the dominant allele is expressed, so the (disease-causing) recessive allele's effect is masked / does not affect the phenotype (1)
-

Question 6 (6 marks)

06.1 [2 marks]

Answer: Family history: 85%; no family history: 17%

- family history: $340 \div 400 \times 100 = 85$ (%) (1)
- no family history: $850 \div 5000 \times 100 = 17$ (%) (1)

Notes: Correct answers with no working score 2 marks.

06.2 [2 marks]

Answer: Family history: 20%; no family history: 4%

- family history: $68 \div 340 \times 100 = 20$ (%) (1)
- no family history: $34 \div 850 \times 100 = 4$ (%) (1)

Notes: Correct answers with no working score 2 marks.

06.3 [2 marks]

Answer: They may be more aware of, and worried about, the risk because a relative is affected; and because the allele is known to be in their family, they are more likely to have inherited it than someone from the general population

- they may be more aware of / more motivated to find out about the risk, since a relative is affected (1)
 - the allele is known to be present in their family, so they are more likely to have inherited it than someone with no family history (1)
-