AQA GCSE Combined Science - Trilogy | 8464

SPECIFICATION POINT 4.6.1.5

Inherited disorders

Mark schemes and examiner comments

Tier	**Foundation**
Question groups	**2**
Available paper years	**2023–2024**

Using this booklet

1. Booklet answer numbers match the question booklet.
2. Use the original question number within marking tables.
3. Examiner comments are included where available.

Name: _____ **Date:** _____

Where a precise marking extract is unavailable, the full source document is included in an appendix. Any missing mark scheme is explicitly identified.

4.6.1.5 Inherited disorders

Answer 01 · Mark scheme

Question 6

Question	Answers	Extra information	Mark	AO / Spec. Ref.
06.1	meiosis	allow phonetic spelling	1	AO1 4.6.1.1 4.6.1.2

Question	Answers	Extra information	Mark	AO / Spec. Ref.
06.2	gametes / eggs / sperm are not (genetically) identical	allow gametes / eggs / sperm are (genetically) different allow cells produced by meiosis are not (genetically) identical	1	AO1 4.6.1.1 4.6.1.2
	(there is) mixing of genetic information / genes / DNA **or** genetic information / genes / DNA from two / both parents	allow genetic information / genes / DNA from each parent ignore gametes fuse unqualified ignore two parents unqualified if no other mark awarded allow **1** mark for mutations	1	

Question	Answers	Extra information	Mark	AO / Spec. Ref.
06.3	heterozygous	ignore dominant / recessive	1	AO2 4.6.1.4

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Answer 01 · Mark scheme

June 2023 | 8464/B/2F | Original Q6

Question	Answers	Extra information	Mark	AO / Spec. Ref.
06.4	(parents gamete genotypes shown) **D d** and **d d**	allow in either position in Punnett square	1	AO2
	(possible offspring genotypes correctly derived) **Dd Dd dd dd**	allow correct derivation of offspring genotypes from incorrect gametes allow 3 correct offspring genotypes for **1** mark	2	AO2
	correct identification of **Dd** offspring as having polydactyly	if derivation not correct and shows **DD** and **Dd**, both must be identified as having polydactyly	1	AO2
	probability must match derived offspring genotypes	if no derivation shown allow 0.5 **or** 50% **or** 1 in 2 **or** 1:1 **or** ½ do **not** accept 1:2 allow correct probability from incorrectly derived offspring	1	AO3 4.6.1.4 4.6.1.5

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Answer 01 · Mark scheme

June 2023 | 8464/B/2F | Original Q6

Question	Answers	Extra information	Mark	AO / Spec. Ref.
06.5	any **two** from: • can find out if the embryo has an (inherited) disorder • can prepare for baby (with the disorder) • can decide whether to continue with the pregnancy • fewer people with (inherited) disorders over time	allow can find out if the embryo has inherited disease ignore can find out if the embryo has disease(s) allow description of preparation for the baby eg access to early treatment allow choice of (in vitro) embryos (to be implanted)	2	AO3 4.6.1.5

Total Question 6	**11**

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Answer 02 · Mark scheme

June 2024 | 8464/B/2F | Original Q1

Question 1

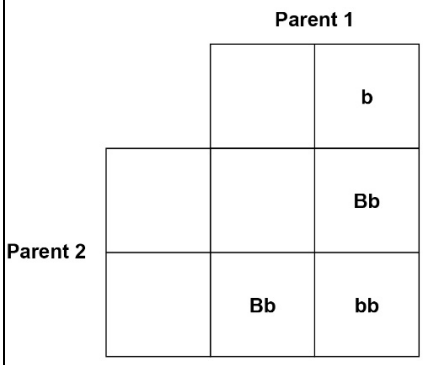
Question	Answers	Extra information	Mark	AO / Spec. Ref.
01.1	cell membrane		1	AO1 4.1.1.2 4.6.1.5

Question	Answers	Extra information	Mark	AO / Spec. Ref.
01.2	dominant		1	AO2 4.6.1.4 4.6.1.5

Question	Answers	Extra information	Mark	AO / Spec. Ref.
01.3	heterozygous		1	AO2 4.6.1.4

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Answer 02 · Mark scheme

Question	Answers	Extra information	Mark	AO / Spec. Ref.
01.4		allow **1** mark for **b** correct allow **2** marks for all 3 offspring genotypes correctly derived from gametes shown / given allow **1** mark for 1 or 2 offspring genotypes correctly derived from gametes shown / given derived genotypes must match parental gamete if given	1 2	AO2 4.6.1.4 4.6.1.5

Question	Answers	Extra information	Mark	AO / Spec. Ref.
01.5	25%	response must match question **01.4** if no answer in question **01.4** allow 25%	1	AO3 4.6.1.4 4.6.1.5

Question	Answers	Extra information	Mark	AO / Spec. Ref.
01.6	screening		1	AO1 4.6.1.5

Question	Answers	Extra information	Mark	AO / Spec. Ref.
01.7	mutation		1	AO1 4.6.2.1

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Answer 02 · Mark scheme

Question	Answers	Extra information	Mark	AO / Spec. Ref.
01.8	any **one** from: • diet • behaviour • infection • named environmental cause of variation such as sunlight	allow descriptions allow examples such as physical damage, scars, tattoos, (body) piercings, smoking, use of hair / nail colourings / dye allow air pollution ignore climate unqualified	1	AO2 4.6.2.1

Total Question 1	**10**

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Answer 01 · Examiner comments June 2023 | 8464/B/2F | Original Q6

Question 6 (Standard demand)

06.1 Phonetic spelling was allowed, and variations of many phonetically correct spellings were seen. Hybrid terms between meiosis and mitosis were not credited. The list principle was applied when students stated both meiosis and mitosis. See page 4 of the mark scheme for an explanation of how lists within responses are marked.

Whilst mitosis was a common error, there were many students who simply put any word that they could relate to sexual reproduction such as fertilisation, ovum, sperm, hormones.

06.2 Few students could give two reasons why sexual reproduction causes variation in the offspring. Marking point 2 was seen more frequently than marking point 1.

Responses that did not gain credit only referred to the offspring having different characteristics, such as eye colour.

For marking point 1 we were looking for the idea of variation in gametes from one parent, or for the variation in gametes more generally, such as 'the genes from the egg and sperm are different'. The word genetically was bracketed and therefore is not required, but could not be contradicted, such as 'the egg and sperm look different'. We cannot put every variation of how students could phrase this in the mark scheme. For example, the mark scheme allowed 'cells produced by meiosis are not identical', so if a student wrote 'cells produced by meiosis are different' that was also worth the mark.

A few students referred to chromosomes crossing over in meiosis. This was a correct response that is beyond the specification and scored marking point 1. There is no expectation for teachers to teach this idea.

We did not credit vague answers that were open to different interpretations such as:

- egg and sperm contain 50% DNA
- half is from mother, half is from father
- genetics is from mother and father
- one gene from each parent.

Errors were seen in some responses that referred to chromosomes. The response needed to be biologically correct to gain the mark. 'They get chromosomes from each parent' or 'Chromosomes mix' or '2 sets of chromosomes mix' was acceptable. However, '2 chromosomes mix' or 'pairs of chromosomes come from each parent' are incorrect. Likewise, '2 genes mix' was incorrect.

There was a fallback mark for 'mutations'. This mark was awarded only if no other mark was given.

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Answer 01 · Examiner comments June 2023 | 8464/B/2F | Original Q6

0 1 . 2 Sexual reproduction produces offspring that are genetically different from each other.

Give **two** reasons why sexual reproduction causes variation in the offspring.

[2 marks]

1 The off spring is a combination of DNA from mother and father

2 The DNA taken from mother and father is not always the same

0 6 . 2 Sexual reproduction produces offspring that are genetically different from each other.

Give **two** reasons why sexual reproduction causes variation in the offspring.

[2 marks]

1 Each sperm and egg cell carry different genes.

2 Mother's and father's ~~are~~ genes are combined differently each time.

The first response (from a Higher tier paper) gave marking point 2 twice, therefore only 1 mark was awarded. The second response (from a Foundation tier paper) gave marking point 1 and marking point 2 for 2 marks.

06.3 This was very well answered. Phonetic spelling was allowed. See page 5 of the mark scheme for information on marking phonetic spelling.

06.4 On the Foundation tier, few students achieved all 5 marks, but partial credit was given frequently, such as correct derivation from incorrect gametes.

On the Higher tier, most students could complete the Punnett square and state the probability of the child having polydactyly. However, few students identified which offspring genotype would result in polydactyly. This could be indicated on any or all offspring, with a circle, line, key or label or any other clear method of identification.

06.5 Clear responses most frequently knew that embryo screening enables us to find out whether an embryo has an inherited disorder. Some confusion was seen between the term disorder and the terms diseases or disabilities.

Many responses were vague, such as 'to see if the embryo is healthy'. Some incorrect responses incorrectly implied that embryo screening is ultrasound screening.

Very few students took the marking point 4 approach of the implications to the population over time.

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Answer 01 · Examiner comments June 2023 | 8464/B/2F | Original Q6

A general misconception was that embryo screening is the same as genetic counselling, so students referred to finding out the likelihood of the embryo having an inherited disorder. The misconception that embryo screening can change DNA or treat inherited disorders was also seen.

Answer 02 · Examiner comments

Question 1 (low demand)

01.1 The vast majority of students could identify that the cell membrane controls the movement of substances into and out of cells.

01.2 More than half of the students could identify that the allele would be dominant.

01.3 More than half of the students correctly selected 'heterozygous' as the term that describes the genotype Bb.

01.4 Most students could interpret the question and use the Punnett square. The Punnet square had been partially completed to reduce the demand of this question. Where students made an error in their choice of gamete for Parent 1, they could still gain the 2 derivation marks using their incorrect gamete. The derivation marks could only be given if the derivation matched the gamete given by the student.

01.5 Students were told that cystic fibrosis is caused by a recessive allele, so the question tested their application of this fact to their Punnett square. The percentage had to match the derivations given in question 01.4. If no derivations were attempted in question 01.4, then an answer of 25% was awarded the mark.

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4.6.1.5 Inherited disorders

Answer 02 · Examiner comments June 2024 | 8464/B/2F | Original Q1

0 1 . 4 Two people plan to have a child.

Both people have the genotype **Bb**.

Complete **Figure 1** to show the possible genotypes of the child. **[3 marks]**

Figure 1

		Parent 1	
		B	b
Parent 2	B	BB	bb
	b	bb	Bb

0 1 . 5 What is the chance that a child of these parents will have cystic fibrosis?

Use **Figure 1**.

[1 mark]

Tick (✓) **one** box.

0% ☐ 25% ☐ 50% ☑ 75% ☐

This student gave the correct Parent 1 gamete, but did not correctly derive any offspring genotypes, therefore 1 mark was awarded for question 01.4.

In question 01.5, the student gave a correct percentage from their Punnett square, therefore the mark was awarded.

01.6 Most students knew that screening was being described.

01.7 Nearly all students knew that mutation is the name of a change in DNA.

01.8 The mark scheme required a cause of environmental variation or the result of the cause. Where a student gave a result of the cause as their response, this was only given credit if it is from an environmental cause. For example, a tattoo would be the result of an environmental cause. However, weight, freckles, skin colour and hair colour are all also affected by genetic causes, so a student needed to qualify these to gain credit. Correct examples commonly seen were 'weight because you eat too much' and 'freckles/skin colour from being out in the sunlight'. Despite the prompt in the question *not* to refer to inheritance or to changes in DNA, these were frequently seen.

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