AQA GCSE Combined Science - Trilogy | 8464

SPECIFICATION POINT 4.6.1.5

Inherited disorders

Mark schemes and examiner comments

Tier	**Higher**
Question groups	**4**
Available paper years	**2020–2024**

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

November 2020 | 8464/B/2H | Original Q5

Question	Answers	Extra information	Mark	AO / Spec. Ref.
05.1	(male gametes) X and Y all offspring correct **Female** X X **Male** **X** **XX** **XX** **Y** **XY** **XY**	 allow correct offspring from incorrect gametes	1 1	AO1 AO2 4.6.1.6
05.2	It is (50%) chance if sperm cells which fertilised the egg has X (or Y) chromosome	allow each / every child / baby / pregnancy has a 50% chance of being male or female (so can be all same sex)	1	AO2 4.6.1.6 4.6.1.4

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Answer 01 · Mark scheme

November 2020 | 8464/B/2H | Original Q5

05.3	both parents shown as heterozygous derivation correct to show 1 unaffected, 2 carriers and 1 affected offspring. **Woman** T t **Man** **T** **TT** **Tt** **t** **Tt** **tt** **tt** identified as offspring with CF correct ratio / probability from their derivations of **tt** e.g. 1:3, 1 in 4, 25%, 0.25	allow correct derivation from incorrect gametes	1 1 1 1	AO2 AO2 AO2 AO3 4.6.1.5 4.6.1.4
05.4	any **three** from: • does not need IVF which is stressful / invasive • higher chance of successful pregnancy as risk of miscarriage is low compared to (30%) success rate of IVF • the test is freely available • the test can be carried out even once the mother is pregnant	allow method 2 is not available to all couples who want it	3	AO3 4.5.3.5 4.6.1.5

4.6.1.5 Inherited disorders

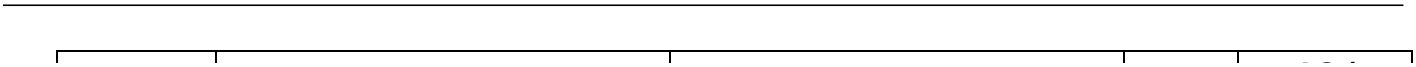
Answer 01 · Mark scheme

05.5	any **one** from: • child will (definitely) not have CF • do not need to consider termination • it is a way to have a child if couples are having fertility problems		1	AO3 4.5.3.5 4.6.1.5
Total			**11**	

4.6.1.5 Inherited disorders

Answer 01 · Mark scheme

November 2020 | 8464/B/2H | Original Q5



4.6.1.5 Inherited disorders

Answer 02 · Mark scheme

November 2021 | 8464/B/2H | Original Q6

Question	Answers	Extra information	Mark	AO / Spec. Ref.
06.1	any **one** from: • (having two) different alleles for a gene / trait / characteristic / disorder • (having) the dominant **and** recessive allele for a gene / trait / characteristic / disorder	ignore examples such as Hh ignore having two different alleles unqualified	1	AO1 4.6.1.4
06.2	father / person 7 **hh** mother / person 8 **Hh**	allow **hh** and **Hh** parental genotypes with each parent unidentified **or** reversed for **1** mark	1 1	AO2 AO2
	(possible offspring correctly derived) **hh** (× 2) **Hh** (× 2)	allow correctly derived offspring from incorrect parental genotype(s)	1	AO2
	(each different phenotype identified) **hh** = has the disorder **Hh** = does not have the disorder	allow from incorrectly derived offspring if incorrectly have **HH** = does not have the disorder	1	AO2
	0.5	allow 50% **or** ½ **or** 1:1 **or** 1 out of 2 **or** 1 in 2 do **not** accept 1:2 allow probability of having disorder correctly derived from incorrect parental genotypes	1	AO3
	(probability of male with disorder) 0.25	allow 25% **or** ¼ **or** 1:3 **or** 1 out of 4 **or** 1 in 4 do **not** accept 1:4 allow probability of male with disorder correctly derived from incorrect probability of having the disorder	1	AO3 4.6.1.4 4.6.1.5 4.6.1.6

4.6.1.5 Inherited disorders

06.3	caused by mutation	allow description, for example change in the genetic code **or** change in base sequence	1	AO2 4.6.2.1 4.6.1.4 4.6.1.5 4.6.1.3
	during meiosis	allow in (germ) cells prior to meiosis allow in (the formation of) gametes / egg / sperm allow during mitosis between fertilisation and birth	1	4.6.1.1 4.6.1.2
	causing a change in amino acid sequence		1	
	causing a different (specific) protein to be produced **or** causing none of a (specific) protein to be produced	causing a different (specific) enzyme to be produced **or** causing none of a (specific) enzyme to be produced allow polydactyly is caused by a dominant allele so if child has one / the allele (with the mutation) they will have the disorder if no other mark awarded allow parents used donated egg / sperm for **1** mark	1	
Total			**11**	

4.6.1.5 Inherited disorders

Answer 03 · Mark scheme

June 2023 | 8464/B/2H | Original Q1

Question 1

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

Question	Answers	Extra information	Mark	AO / Spec. Ref.
01.2	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.
01.3	heterozygous	ignore dominant / recessive	1	AO2 4.6.1.4

4.6.1.5 Inherited disorders

Answer 03 · Mark scheme

Question	Answers	Extra information	Mark	AO / Spec. Ref.
01.4	(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

4.6.1.5 Inherited disorders

Answer 03 · Mark scheme

Question	Answers	Extra information	Mark	AO / Spec. Ref.
01.5	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 1	11

4.6.1.5 Inherited disorders

Answer 04 · Mark scheme

June 2024 | 8464/B/2H | Original Q3

Question 3

Question	Answers	Extra information	Mark	AO / Spec. Ref.
03.1	parents of child with AKU / disorder do not have AKU / disorder **or** child(ren) with AKU / disorder have parents who do not	allow **A** and **B** do not have AKU, but child / **D** does (have AKU) allow **F** and **G** do not have AKU, but child / children / **K** / **L** does (have AKU) allow child(ren) with AKU / disorder have parents who are carriers	1	AO3 4.6.1.4

Question	Answers	Extra information	Mark	AO / Spec. Ref.
03.2	G		1	AO2 4.6.1.4

4.6.1.5 Inherited disorders

Answer 04 · Mark scheme

Question	Answers	Extra information	Mark	AO / Spec. Ref.
03.3	(parental gametes) (female) **a and a** (male) **A and a**	allow **1** mark for **a a and A a** parental gametes reversed	1 1	AO2 AO2
	offspring correctly derived **Aa Aa aa aa**	allow correctly derived offspring from incorrect parental gametes	1	AO2
	offspring phenotype correctly derived **Aa** = no AKU **aa** = AKU	allow correctly derived phenotypes from incorrect parental gametes or incorrect offspring genotypes allow inclusion of **AA** = no AKU, if their cross derives this genotype ignore reference to carriers	1	AO2
	(probability =) 0.5	allow 50% **or** ½ **or** 1 in 2 **or** 1:1 **or** 50:50 probability must match offspring genotype do **not** accept 1:2 **or** 50/50	1	AO3 4.6.1.4

Question	Answers	Extra information	Mark	AO / Spec. Ref.
03.4	(mutation causes) incorrect sequence of amino acids		1	AO2 4.6.1.3
	(which) causes the enzyme to be a different shape	allow (which) causes the active site to be a different shape allow which means the enzyme and substrate cannot bind ignore the enzyme is denatured do **not** accept no enzyme produced	1	

Question	Answers	Extra information	Mark	AO / Spec. Ref.
03.5	FSH **and** LH given / injected (into female) to stimulate maturation of egg(s)		1	AO1 4.5.3.3 4.5.3.5
	(collected) egg(s) are fertilised (in laboratory)	allow (collected) egg(s) fused with sperm (in laboratory) allow sperm is injected into egg (in laboratory)	1	
	fertilised egg undergoes mitosis	allow cell(s) undergo mitosis allow fertilised egg divides / develops to form embryo	1	
	(one or two) embryo(s) inserted into uterus / womb **or** (one or two) ball(s) of cells inserted into uterus / womb		1	

Question	Answers	Extra information	Mark	AO / Spec. Ref.
03.6	any **two** from: • risk to embryo	allow risk of miscarriage do **not** accept risk to embryo due to radiation	2	AO3 4.6.1.5
	• risk to female / mother • stressful process • may lead to termination of the pregnancy	allow may lead to destruction of the embryo		
	• (high) cost (of the screening process) • embryo cannot give consent	allow idea of increased prejudice against other people with (inherited) disorders ignore references to religion / ethics		

Total Question 3	15

4.6.1.5 Inherited disorders

Answer 02 · Examiner comments November 2021 | 8464/B/2H | Original Q6

Question 6 (standard/high and high demand)

Few students could define the term heterozygous. Some insufficient responses gave examples such as Hh, but did not answer the question. Confusion was seen between the terms allele, gene and chromosome.

In **06.2** there were clear instructions in the question regarding the level of detail required. Many students only attempted a Punnett Square, not identifying the genotypes of the parents and the phenotype of each offspring. **06.3** was beyond the understanding of most students. Common incorrect answers referred to polydactyly skipping a generation or the parents being carriers, despite the detail given in the question.

4.6.1.5 Inherited disorders

Concluding remarks

It was clear that in the unusual teaching circumstances leading up to this exam had made it not possible for some topics and practical work to be covered in the normal way. It was a small and atypical cohort of students who have chosen to take the autumn series of examinations. Gaps in both knowledge and skills were evident.

4.6.1.5 Inherited disorders

Mark Ranges and Award of Grades

Grade boundaries and cumulative percentage grades are available on the Results Statistics page of the AQA Website.

4.6.1.5 Inherited disorders

Answer 03 · Examiner comments

Question 1 (Standard demand)

01.1 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 the 'Information for examiners' on 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.

01.2 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, 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.

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 fall back mark for 'mutations'. This mark was awarded only if no other mark was given.

4.6.1.5 Inherited disorders

Answer 03 · Examiner comments June 2023 | 8464/B/2H | Original Q1

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.

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

01.4 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.

01.5 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.

4.6.1.5 Inherited disorders

Answer 03 · Examiner comments June 2023 | 8464/B/2H | Original Q1

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 04 · Examiner comments

Question 3 (standard, standard/high & high demand)

03.1 There was only 1 mark available here and for this to be awarded it was essential that reference was made to both parents (without the disorder) and their children (who have the disorder). A common misconception was for students to base their descriptions on the (low) frequency of AKU within the family.

Students should be aware that using 'it' will be treated in terms of how the question is phrased, unless the student clarifies what they mean by 'it'. Please see page 4 of the mark scheme for more guidance on the use of 'it'. Here, 'it' unqualified was considered equivalent to 'AKU'.

No evidence could be obtained from C and D as parents. Answers which described this part of the family tree as evidence gained no credit as it is incorrect biology, even if a student went on to make further correct points.

Examples of answers which were too vague to match the mark scheme included:

- 'D has AKU and this means that his parents are heterozygous' ('Heterozygous' does not clearly suggest that a recessive AKU allele is present but the parents do not have AKU)
- 'A and B don't have AKU but future generations do' ('Future generations' is too vague)
- 'AKU skips generations'.

03.2 Fewer than 50% of students could identify which person is definitely heterozygous for AKU. More students selected the incorrect answer of 'D'.

03.3 Most students were able to correctly identify the gametes for both parents, although where one of these marks was not awarded it was often because students presumed that the female with AKU had the genotype AA rather than aa and therefore were struggling to tell the difference between recessive and dominant despite being told AKU was recessive earlier in the question.

Almost all students were able to derive the offspring in the Punnett square and were awarded marking point 3.

For marking point 4, very few students attempted to identify the offspring phenotypes, and when there was an attempt at this, the mark wasn't awarded if only the phenotype 'having AKU' was indicated rather than the phenotype for both having AKU and not having AKU. Many students drew circles around offspring genotypes in the belief that this would be sufficient to suggest a phenotype of 'having AKU', but this was not creditworthy. Another frequent mistake was referring to the phenotype of Aa offspring as being 'heterozygous' or 'a carrier'. Although not incorrect descriptions, these are not actual phenotypes.

Giving the probability of having AKU was, in the majority of cases, done correctly and was usually given as a percentage or a fraction. This was very rarely given as a ratio. When incorrect gametes were given, a common mistake made when working out the probability was that offspring with Aa or AA would have AKU.

Other, more occasional, errors included giving an incorrect probability, such as 1:2 or 50/50 or 50 and using symbols other than those (A and a) directed in the question.

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4.6.1.5 Inherited disorders

Answer 04 · Examiner comments June 2024 | 8464/B/2H | Original Q3

It was noted that quite a few students changed their minds when answering this question and made alterations to the gametes and genotypes they originally opted for. If students do this, they must make sure that the changes are unambiguous and, if necessary, rewrite their choices. It was sometimes very difficult for examiners to decide what the student intended to be marked.

0 3 . 3 A female who has AKU and a male who is heterozygous for AKU plan to have a child.

Determine the probability that the child will have AKU.

You should:

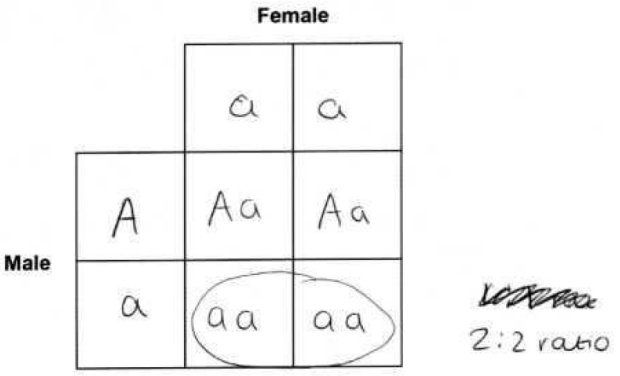
- complete **Figure 4**
- identify the phenotype of each offspring genotype
- use the symbols:

  A = dominant allele

  a = recessive allele.

[5 marks]

Figure 4



Probability that the child will have AKU = 50%

In a typical response, as shown above, students gave the correct parental gametes, and derived offspring genotypes correctly. The student has not indicated the phenotype of each offspring genotype, therefore marking point 4 was not awarded. However, the probability is acceptable, so 4 marks could be awarded.

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4.6.1.5 Inherited disorders

Answer 04 · Examiner comments June 2024 | 8464/B/2H | Original Q3

03.4 Students found this question difficult with very few gaining both marks. They struggled to make appropriate connections between the mutation and the non-functioning enzyme.

Answers were often too general and repetitive of information given in the question to be adding any value, such as 'a mutation could change the cell's DNA to stop producing functioning enzymes' or 'if the gene has changed it will mean that the enzyme has changed'.

In other cases, students clung to the familiarity of terms in the question but created their own, inappropriate, narratives around them. So, 'mutation' triggered reference to 'random changes', incidence in the family tree, links to cancer and 'uncontrollable cell division'; 'protein' prompted discussions of ribosomes, protein digestion by protease, proteins being needed for growth and repair; 'enzyme' opened the way to details of denaturation.

Misconceptions were quite frequently seen, such as:

- 'DNA is made of amino acids'
- 'DNA a protein'
- 'genes make amino acids that code for a protein'
- 'the enzyme is 'mutated'
- 'the enzyme changes shape and can then no longer fit the active site'.

Overall, marking point 2 was seen far more frequently than marking point 1. Students clearly felt more comfortable discussing why enzymes might not be functioning rather than the reason behind this (the immediate consequence of the mutation).

03.5 This question was set at high demand. It closely reflects the specification content for the use of hormones in the treatment of infertility. Answers were often confused and poorly sequenced. Appreciation of the distinctions between 'egg', 'fertilised egg' and 'embryo' and between 'ovary' and 'uterus' was often lacking.

For the first mark, students had to refer only to FSH and LH being given to the female to stimulate maturation of eggs. Incorrect answers included ideas of the hormones 'increasing the chances of fertilisation', 'making the eggs more fertile', 'strengthening the eggs'. Students occasionally suggested that FSH and LH production was stimulated through treatment rather than the hormones being given directly. No credit was given for this.

The second marking point was the most commonly awarded mark. Credit was not given, however, for vague references such as the eggs and sperm were 'combined' or 'joined together'. The term 'fertilisation'/'fusion' was required or a suitable description of sperm being injected into eggs (ICSI). No mark was given when the student described fertilisation 'in the woman'.

Very few students achieved marking point 3. Words that were too vague such as 'growing'/'forming'/'turning into' were used instead. Mention of 'mitosis' was exceptionally rare. No credit was given if it was implied that embryo development only happened at a later stage within the uterus.

For the fourth mark, many students did not state that the embryo was inserted into the uterus. Answers which said that the embryo was 'put back into the woman' were insufficient. Reference to anything other than the 'embryo' or the 'ball of cells' at this stage, such as 'fertilised egg'/'baby', was incorrect. Some students wrongly suggested that the embryos were inserted into ovaries.

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4.6.1.5 Inherited disorders

Answer 04 · Examiner comments June 2024 | 8464/B/2H | Original Q3

03.6 This was generally well answered. The most commonly awarded marks were for bullet point 4 and bullet point 5, as students clearly understood the high cost of the process and that it might lead to parents choosing to terminate their pregnancy. Some students attempted bullet point 1 but did not understand the process and wrote about the embryo being harmed due to the radiation used in the process, which was incorrect.

Some students simply wrote 'ethics' without giving details on why some people might think this and there were often references to 'designer babies', which was incorrect. Some students appeared to be talking about IVF, such as 'not always successful', with some confusion about what embryo screening involves.

Some students described ideas of prejudice against people with inherited disorders. This was described in a variety of ways and was creditworthy.