AQA GCSE Biology | 8461

SPECIFICATION POINT 4.6.1.7

Inherited disorders

Mark schemes and examiner comments

Tier	**Foundation**
Question groups	**11**
Available paper years	**2008–2024**

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.

4.6.1.7 Inherited disorders

Answer 01 · Mark scheme June 2008 | BLY2F | Original Q8 | Legacy

question	answers	extra information	mark
8(a)	cell membranes		1
8(b)(i)	two recessive / cystic fibrosis / faulty / diseased / the allele(s) / genes from Bob **and** Carol / both parents / the parents	two can be implied by second marking point ignore chromosomes if no other marks awarded 'Carol is a carrier' gains **1** mark	1 1
8(b)(ii)	(inherited) dominant / normal allele / gene from Carol / mother	ignore references to recessive allele / gene from father / Bob if no other marks awarded he has <u>just</u> / <u>only</u> one recessive allele gains **1** mark	1 1
8(c)(i)	reduce number of people with cystic fibrosis (in population) **or** reduce health-care costs **or** expensive to have baby with cystic fibrosis	accept to allow decision / emotional argument qualified eg allows abortion **or** allows people to make choices about termination **or** help to prepare financially / emotionally etc	1

Question 8 continued on next page…

4.6.1.7 Inherited disorders

Answer 01 · Mark scheme

COMPONENT NUMBER: BLY2F

COMPONENT NAME: Additional Science / Biology

STATUS: Final

DATE: June 2008

Question 8 continued…

question	answers	extra information	mark
8(c)(ii)	any **one** from: • possible damage / risk to embryo / fetus / baby • screening / it is expensive • (may) have to make ethical / moral / religious decisions • right to life	 allow possible harm / risk to mother ignore not natural / playing God / unethical / immoral / religious unqualified	1
Total			**7**

4.6.1.7 Inherited disorders

Answer 02 · Mark scheme

Question 2

question	answers	extra information	mark
2(a)(i)	sex cells		1
2(a)(ii)	chromosomes		1
2(b)(i)	two		1
2(b)(ii)	recessive		1
2(c)(i)	cell membrane	allow membrane	1
2(c)(ii)	cytoplasm		1
2(d)(i)	A		1
2(d)(ii)	B		1
Total			**8**

4.6.1.7 Inherited disorders

Answer 02 · Mark scheme

January 2010 | BLY2F | Original Q2 | Legacy

BLY2F

4.6.1.7 Inherited disorders

Answer 03 · Mark scheme

June 2019 | BLY2F | Original Q4 | Legacy

Question 4

question	answers	extra information	mark
4(a)(i)	circle unshaded	mark independently could be in body of script	1 1
4(a)(ii)	(Harriet) dd DD Dd	in first box if another letter is chosen it must be used throughout and upper or lower case must be clear	1 1 1
4(b)(i)	to check for the D allele.		1
4(b)(ii)	any **one** from: • may harm / kill foetus / embryo / baby / mother • immoral / unethical / religion	allow could affect the baby ignore playing God ignore references to unnatural ignore wrong unqualified ignore expense / prejudice unqualified ignore lack of permission ignore results are unreliable	1
Total			**7**

4.6.1.7 Inherited disorders

Answer 03 · Mark scheme June 2010 | BLY2F | Original Q4 | Legacy

BLY2F

Answer 04 · Mark scheme June 2011 | BLY2F | Original Q3 | Legacy

Question 3

question	answers	extra information	mark
3(a)	insulin	extra ring drawn cancels the mark	1
3(b)	pancreas	extra ring drawn cancels the mark	1
3(c)	diabetes	extra ring drawn cancels the mark	1
Total			3

Answer 04 · Mark scheme June 2011 | BLY2F | Original Q3 | Legacy

BLY2F

4.6.1.7 Inherited disorders

Answer 05 · Mark scheme

June 2012 | BL2FP | Original Q7 | Legacy

Question 7

question	answers	extra information	mark
7(a)(i)	recessive allele		1
7(a)(ii)	carriers		1
7(b)(i)	6	allow nn	1
7(b)(ii)	1 in 4 / 0.25 / ¼ / 25% / 1:3	do **not** accept '3:1' / 1:4 / 1 in 3 / 25	1
7(c)	**advantage:** detect CF qualified – eg at early stage / before becoming pregnant **or** (only) healthy children produced **disadvantage:** some embryos are destroyed / may damage embryo	 allow 'after only 3 days' allow reduces health care costs allow increased risk of miscarriage ignore not natural ignore cost	 1 1
Total			6

4.6.1.7 Inherited disorders

Answer 05 · Mark scheme

June 2012 | BL2FP | Original Q7 | Legacy

BL2FP

4.6.1.7 Inherited disorders

Answer 06 · Mark scheme

June 2013 | BL2FP | Original Q5 | Legacy

Question 5

question	answers	extra information	mark
5(a)(i)	1		1
5(a)(ii)	fertilisation / fusion	allow sexual reproduction allow fertilise / fuse ignore joining	1
5(b)(i)	**Dd**		1
5(b)(ii)	**dd**		1
5(c)(i)	1 in 2		1
5(c)(ii)	0		1
Total			**6**

4.6.1.7 Inherited disorders

Answer 07 · Mark scheme

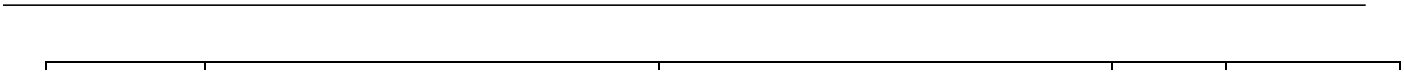
June 2015 | BL2FP | Original Q1 | Legacy

Question	Answers	Extra information	Mark	AO / Spec. Ref.
1(a)	**A** = nucleus **B** = (cell) membrane	allow phonetic spelling	1 1	AO1 2.1.1a, d
1(b)	for repair / growth **or** to replace cells	ignore new cells / skin	1	AO1 2.7.1j
1(c)(i)	embryos		1	AO1 2.7.1k
1(c)(ii)	paralysis		1	AO1 2.7.1l,m
Total			**5**	

4.6.1.7 Inherited disorders

Answer 07 · Mark scheme

June 2015 | BL2FP | Original Q1 | Legacy



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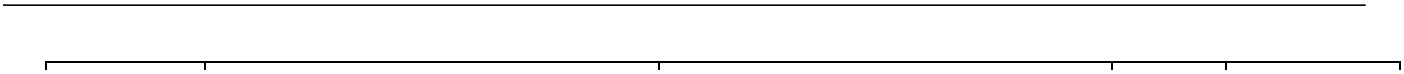
Answer 08 · Mark scheme June 2015 | BL2FP | Original Q5 | Legacy

Question	Answers	Extra information	Mark	AO / Spec. Ref.
5(a)(i)	(female) has XX / only X's / no Y	allow has X chromosomes ignore ref to genes / cells	1	AO2 2.7.2b, 2.7.3a
5(a)(ii)	extra chromosome / has 47 chromosomes / one set has 3 copies no. 18	ignore reference to chromosome numbers other than 47 or no. 18	1 1	AO2 2.7.1a,e, 2.7.3a
5(b)(i)	14	allow in range of 13.5 to 14.5	1	AO2 2.7.1a,e, 2.7.3a
5(b)(ii)	7	allow in range of 6.75 to 7.25 accept ecf from 5bi	1	AO2 2.7.1a,e, 2.7.3a
5(c)	Advantages: any **two** from: • more than 1 embryo (so more chance of success) • tested at 3 days cf 10 weeks **or** tested earlier • tested before pregnancy • no termination / abortion • spare embryos have a potential use Disadvantages: any **one** from: • needs an operation • (spare) embryos / human life destroyed / harmed • higher cost • embryos might not implant / might not develop	allow method 2 may cause a miscarriage tested when only 3 days old accept described hazard of operation must be comparative	2 1	AO3 2.7, 2.7.3d
Total			**8**	

4.6.1.7 Inherited disorders

Answer 08 · Mark scheme

June 2015 | BL2FP | Original Q5 | Legacy



4.6.1.7 Inherited disorders

Answer 09 · Mark scheme June 2015 | BL2FP | Original Q9 | Legacy

Question	Answers	Extra information	Mark	AO / Spec. Ref.
9(a)(i)	in the chromosome(s) in the nucleus	ignore genes / alleles allow nuclei allow mitochondria	1 1	AO1 2.7.2f, 2.7.1b
9(a)(ii)	the DNA / chromosomes / genes are replicated / copied / multiplied / doubled / duplicated	allow DNA is cloned ignore same DNA / chromosomes / genes if unqualified	1	AO1 2.7.1a,c,n
9(b)(i)	1 / one		1	AO2 2.7.2c,e, 2.7.3a,c
9(b)(ii)	2 / two		1	AO2 2.7.2c,d, 2.7.3a,b
9(c)	**B**		1	AO3 2.7.2i
Total			**6**	

4.6.1.7 Inherited disorders

Answer 10 · Mark scheme

November 2020 | 8461/2F | Original Q3

Question	Answers	Extra information	Mark	AO / Spec. Ref.
03.1	4 / four		1	AO1 4.6.1.2
03.2	23 / twenty three	do **not** accept 23 pairs	1	AO1 4.6.1.2
03.3	a different form of a gene		1	AO1 4.6.1.6
03.4	heterozygous		1	AO2 4.6.1.6
03.5 mark with 3.6 and 3.7		allow 2 correct for **1** mark	2	AO2 4.6.1.1 4.6.1.2 4.6.1.6
03.6 mark with 3.5 and 3.7	ring around any **Dd**	allow ecf from question **03.5**	1	AO2 4.6.1.6
03.7 mark with 3.5 and 3.6	percentage must match answer given to questions **03.5** and **03.6**	if no answer in question **03.5** allow 50 %	1	AO3 4.6.1.6
03.8	mutation / mutated	do **not** accept mutant	1	AO1 4.6.2.1

4.6.1.7 Inherited disorders

Answer 10 · Mark scheme

November 2020 | 8461/2F | Original Q3

03.9	any **one** from: • to help them prepare • to inform whether to consider having an abortion • to find out if they have passed on the disorder	allow to see if the child / embryo has the disorder allow answers referring to genetic disorders, or specific example such as Dupuytren's / cystic fibrosis	1	AO3 4.6.1.7
Total			**10**	

4.6.1.7 Inherited disorders

Answer 10 · Mark scheme

November 2020 | 8461/2F | Original Q3

4.6.1.7 Inherited disorders

Answer 11 · Mark scheme

June 2024 | 8461/1F | Original Q8

Question 8

Question	Answers	Extra information	Mark	AO / Spec Ref.
08.1	nucleus	allow chromosome ignore in the DNA	1	AO1 4.1.1.2 4.1.2.1

Question	Answers	Extra information	Mark	AO / Spec Ref.
08.2	**A**, **D** and **E**		1	AO1 4.2.2.1 4.2.2.2

4.6.1.7 Inherited disorders

Answer 11 · Mark scheme

Question	Answers	Mark	AO / Spec Ref.
08.3	**Level 3:** Relevant points (reasons / causes) are identified, given in detail and logically linked to form a clear account.	5–6	AO2
	Level 2: Relevant points (reasons / causes) are identified, and there are attempts at logical linking. The resulting account is not fully clear.	3–4	AO2
	Level 1: Points are identified and stated simply, but their relevance is not clear and there is no attempt at logical linking.	1–2	AO1
	No relevant content.	0	4.2.2.1 4.4.2.1 4.4.2.3
	Indicative content: **Difficulty digesting food** • less / no lipase • (so) less / no fat broken down o into fatty acids o into glycerol • less / no carbohydrase / amylase • (so) less / no carbohydrate / starch broken down o into glucose / sugar • less / no protease • (so) less / no protein broken down o into amino acids **Difficulty gaining body mass** • less / no absorption o of small / soluble molecules o of fatty acids o of glycerol o of glucose / sugar o of amino acids • fewer molecules **or** fewer amino acids available for building protein / muscle / cells / tissues • less fat stored • less respiration • less energy • (so less energy) for building new molecules / cells / tissues For **Level 3** details of difficulty digesting food **and** difficulty gaining body mass are needed.		

4.6.1.7 Inherited disorders

Answer 11 · Mark scheme

Question	Answers	Extra information	Mark	AO / Spec Ref.
08.4	large surface / area	allow large surface / area to volume (ratio)	1	AO1 4.1.3.1 4.2.2.2
	(large) capillary network **or** good / efficient blood supply	allow many capillaries	1	
	walls are thin **or** walls are one cell thick	ignore references to membranes ignore alveoli are thin ignore alveoli are one cell thick do **not** accept thin cell walls ignore references to alveoli being moist ignore steep concentration gradient	1	

4.6.1.7 Inherited disorders

Answer 11 · Mark scheme

Question	Answers	Extra information	Mark	AO / Spec Ref.
08.5	less (aerobic) respiration	allow (more) anaerobic respiration	1	AO2 4.4.2.1 4.4.2.2 4.4.2.3
	(so) less energy (released)	do **not** accept less energy produced / made / created	1	
	(results in) less muscle contraction **or** (results in) reduced metabolism **or** (results in) increased breathing rate / depth **or** (results in) increased heart rate	allow relevant named metabolic processes allow (results in) person getting out of breath	1	
	OR (more) anaerobic respiration (1) (so) lactic acid produced (1)			
	(results in) <u>muscle</u> fatigue **or** (results in) less <u>muscle</u> contraction **or** (results in) increased breathing rate / depth **or** (results in) increased heart rate (1)	allow <u>muscle</u> ache / cramp / tiredness / pain allow (results in) person getting out of breath		

Total Question 8	14

4.6.1.7 Inherited disorders

Answer 10 · Examiner comments November 2020 | 8461/2F | Original Q3

Question 3 (low & standard demand)

This question was about meiosis, chromosomes and an inherited disorder in humans. A little over one-third of students gave the correct answer of '4' cells being formed by meiosis in **03.1**. There were many other alternatives offered: '2' was, not unexpectedly, a common incorrect response and other suggestions included '8' and as many as '50 million' as these last students presumably either misread the question or misunderstood the process of meiosis.

For **03.6** most students ringed an appropriate offspring genotype although a significant minority (about 1 in 15) did not answer this question. The most likely reason for this is that students missed the question, rather than not understanding what they had to do. The lack of answer lines, spaces or boxes to tick should not be seen by students to mean that nothing needs to be done. Students should be reminded to read all of the words on the paper and respond accordingly. Those students who gave incorrect responses often ringed contradictory genotypes, showing a lack of understanding of genetic principles.

4.6.1.7 Inherited disorders

Answer 11 · Examiner comments

June 2024 | 8461/1F | Original Q8

Question 8 (standard demand)

08.1 57% of students gained credit in this question.

08.2 88% of students selected the correct answer in this question.

08.3 This extended response question was not answered well. Most students found it very difficult to access and the majority of responses simply restated the information given in the question or detailed everything the student knew about cystic fibrosis (CF) and the lungs. Less than 13% of students achieved anything more than one mark, with 57% achieving zero.

Students achieving Level 1 gave one or more simple, unconnected, but appropriate statements which reflected the difficulties encountered. To enter Level 2, there had to be at least the attempted linking of one idea with another, which reflected either the difficulty in digesting food or the difficulty in gaining body mass. To enter Level 3 students had to show at least some attempted linking of two or more ideas and they had to address, even if only minimally, both difficulties in digesting food and gaining body mass.

Responses invariably focused only on difficulties with digestion, as the context of this topic probably seemed more familiar. General comments which simply reworded what had been given in the question such as 'people with CF have fewer enzymes so will find it hard to breakdown food' or 'the gut will find it difficult to function' were repeated time and again in paragraphs but added nothing of value to answers. Some students gravitated towards the familiarity of 'enzymes' and proceeded to give unnecessary details of enzyme action in general and of possible denaturation. Others followed on from the mention of CF by inappropriately referring to breathing difficulties or by suggesting that excess mucus might 'block the gut'. When ideas were successfully linked, these were most often in relation to individual digestive enzymes and their substrates. Occasional additional points were given such as 'reduced absorption' or 'less fat could be stored' but these were not often considered at this level.

The statements students make in these extended response questions do not need to be long or complex and it is never necessary to include all the points listed in the indicative content on the mark scheme to achieve the highest level. Neither do responses have to be written in complete sentences - succinct bullet points often suffice and may even help students to organise their ideas more effectively. As is always the case with extended response questions, however, answers are considered holistically and always in a positive framework.

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4.6.1.7 Inherited disorders

Answer 11 · Examiner comments June 2024 | 8461/1F | Original Q8

08.4 3% of students achieved full marks in this question, with 13% achieving two marks and 31% achieving one mark. This question asked for a description of the features of the (individual) alveoli that help maximise gas exchange rather than the lungs in general. Most students struggled to describe three features, often missing out on marks due to their responses lacking specificity. An answer such as they are 'one cell thick' or 'thin' had to be related to the alveolar walls to be given credit. Some students referred to there being a 'short diffusion distance' but as this is a consequence of the thin walls and not a described feature, they could not gain the mark. Students should be reminded to double check the command word given in each question; explanatory detail in 'describe' questions is not required, nor creditworthy.

08.5 Fewer than 20% of students scored any credit in this question, despite many making lengthy attempts. Answers were usually too general and invariably did not offer any clear explanations. Two routes were available in the mark scheme – one which reflected there being less oxygen for aerobic respiration and the other which reflected the inevitable increase in anaerobic respiration. The variety of answers which did not gain credit included vague suggestions such as 'tiredness', it being 'hard to breathe', the 'heart rate is affected' or 'weak muscles' as well as incorrect ones such as 'blood stops flowing' or there is 'no respiration'.

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4.6.1.7 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.

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