AQA GCSE Biology | 8461

SPECIFICATION POINT 4.6.1.7

Inherited disorders

Mark schemes and examiner comments

Tier	**Higher**
Question groups	**13**
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 | BLY2H | Original Q2 | Legacy

question	answers	extra information	mark
2(a)	cell membranes		1
2(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
2(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
2(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 2 continued on next page…

4.6.1.7 Inherited disorders

COMPONENT NUMBER: BLY2H

COMPONENT NAME: Additional Science / Biology

STATUS: Final

DATE: June 2008

Question 2 continued…

question	answers	extra information	mark
2(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 01 · Mark scheme

June 2008 | BLY2H | Original Q2 | Legacy

COMPONENT NUMBER: BLY2H

COMPONENT NAME: Additional Science / Biology

STATUS: Final

DATE: June 2008

4.6.1.7 Inherited disorders

Answer 02 · Mark scheme

January 2009 | BLY2H | Original Q6 | Legacy

question	answers	extra information	mark
6(a)	1 in 4 / $^{1}/_{4}$ / 1: 3 / 25% / 0.25	do **not** accept 3:1 / 1:4 / 2:6	1
6(b)	**either** from C **and** D	accept synonyms for dominant / recessive eg. Normal / faulty accept genetic diagram if clearly referring to correct individuals or genotypes on family tree allow 'gene' for 'allele'	
	any **three** from: • C **and** D have disorder • I/J don't have disorder • C **and** D have dominant **and** recessive alleles • recessive alleles from C **and** D passed to I/J **or** I/J have two recessive alleles	ignore 'C & D are carriers' NB if allele was recessive then all offspring of C **and** D would have the disorder = **3** marks	3
	or from A **and** B	assume response refers to A+B unless contradicted	
	• A is homozygous recessive / rr, **and** B is heterozygous / Rr can be shown in words or symbols • offspring can be rr **or** Rr described	allow any symbol allow without key	
6(c)(i)	(embryos) checked for inherited / genetic disorders / conditions	accept diseases for disorders	1

Question 6 continued on next page…

COMPONENT NUMBER: BLY2H

COMPONENT NAME: Additional Science / Biology

STATUS: Final

DATE: January 2009

Question 6 continued…

question	answers	extra information	mark
6(c)(ii)	any **three** from: • C/D have disorder / have dominant allele • chance of embryo / foetus / child having disorder **or** may pass on alleles for disorder to their offspring • C/D might want to decide on termination **or** prepare for child with disorder • G **and** H don't have disorder / both homozygous recessive / have no dominant alleles (for this disorder) • so offspring (of G **and** H) cannot / don't have disorder	accept disease / condition accept 'gene' for 'allele' ignore reference to 'carriers'	3
Total			**8**

4.6.1.7 Inherited disorders

Answer 03 · Mark scheme June 2009 | BLY2H | Original Q6 | Legacy

Question 6

question	answers	extra information	mark
6(a)	(genotype / gametes from **T** / mother) h and h (accept h) (genotype / gametes from **U** / father) H and h offspring genotypes: correctly derived from correct gametes chance = 50% / ½ / 0 .5 / 1 : 1 / 1 out of 2	eg may be in punnett square allow own upper and lower case symbols or allow any symbol correctly used with key do **not** accept 1 : 2 allow irrespective of working	1 1 1 1
6(b)(i)	(cells from) embryo tested for particular allele / gene / (named) genetic condition / the disease / gender	ignore x-rays / ultra sound	1
6(b)(ii)	any **one** from: • possible harm to embryo **or** mother • sensible reference to abortion • hard to make moral / ethical decisions	ignore moral / ethical unqualified ignore not natural / playing God references to other ideas eg designer babies must be well argued	1
6(c)(i)	(cystic fibrosis is) recessive (allele / gene) need two alleles / genes **or** need one allele / gene from each parent **or** both parents must have cystic fibrosis allele / gene	allow not dominant	1 1

Question 6 continues on the next page…

BLY2H

Question 6 Continued

question	answers	extra information	mark
6(c)(ii)	any **three** from: • osmosis / diffusion • more concentrated solution outside cells / in mucus • water moves from dilute to more concentrated solution • partially permeable membrane (of cell)	do **not** accept reference to movement of ions / solution by osmosis / diffusion assume concentration is concentration of solute unless they tell us otherwise allow correct references to movement of water in relation to concentration gradient	3
6(c)(iii)	ribosomes		1
Total			**12**

4.6.1.7 Inherited disorders

Answer 04 · Mark scheme

Question 5

question	answers	extra information	mark
5(a)	both parents **Aa**	accept other upper and lower case letters without key **or** symbols with a key allow shown as gametes in punnet square	1
	aa in offspring correctly derived from parents / **aa** correctly derived from the parents given	ignore other offspring / gametes for this mark parents do not have to be correct	1
	offspring **aa** identified as having cystic fibrosis	may be the only offspring shown **or** circled / highlighted / described	1
5(b)(i)	any **one** from: • sure / greater chance of healthy / non-cystic fibrosis egg / embryo / child • greater chance of fertilisation	accept converse if clear eg if you (only) took one it might have cystic fibrosis / might not be fertilised accept some may have the allele reference to suitable embryo is insufficient	1

Question 5 continues on the next page

BLY2H

Question 5 continued

question	answers	extra information	mark
5(b)(ii)	**advantages** any **two** from • greater / certain chance of having child / embryo without cystic fibrosis / healthy • child with cystic fibrosis difficult / expensive to bring up • cystic fibrosis (gene / allele) not passed on through generations **disadvantages** any **two** from: • operation dangers eg infection • ethical or religious issues linked to killing embryos • (high) cost • possible damage to embryo (during testing for cystic fibrosis / during operation) **plus** **conclusion** a statement that implies a valued, qualified judgement eg it is right because the risk of infection is small **or** eg it is wrong because embryos are killed	**to gain 3 marks both advantages <u>and</u> disadvantages must be given** ignore references to abortion unless qualified by later screening ignore risk unqualified accept wrong / cruel to kill embryos accept right to life **Note:** the conclusion mark cannot be given unless a reasonable attempt to give both an advantage and a disadvantage has (already) been made do **not** award the mark if the conclusion only states that advantages outweigh disadvantages	max 3 1
Total			**8**

4.6.1.7 Inherited disorders

Answer 04 · Mark scheme

January 2010 | BLY2H | Original Q5 | Legacy

BLY2H

4.6.1.7 Inherited disorders

Answer 05 · Mark scheme

Question 6

question	answers	extra information	mark
6(a)	cystic fibrosis (allele / gene) recessive carrier has only one cystic fibrosis allele / gene	allow an annotated genetic diagram accept carrier is heterozygous accept any symbol with key or accept conventional use of symbols penalise use of chromosome once only	1 1
6(b)	any **one** from: • Huntington's (allele / gene) dominant • (to have Huntington's) need only one Huntington's allele / gene		1
Total			**3**

4.6.1.7 Inherited disorders

Answer 05 · Mark scheme

January 2011 | BLY2H | Original Q6 | Legacy

BLY2H

4.6.1.7 Inherited disorders

Answer 06 · Mark scheme

Question 7

question	answers	extra information	mark
7(a)(i)	(alternative) forms / types of a / the same gene		1
7(a)(ii)	only expressed if 2 copies inherited **or** not expressed if other allele present	allow over ruled / over powered by the other allele	1
7(b)(i)	**Nn**	ignore heterozygous	1
7(b)(ii)	genetic diagram including:	accept alternative symbols, if defined	
	gametes: **N** and **n** from both parents	accept alternative symbols if correct for answer to (b)(i)	1
	correct derivation of offspring genotypes: **NN Nn Nn nn**	allow if correct for candidate's parental genotypes / gametes	1
	identification of **nn** as having cystic fibrosis		1

Question 7 continues on the next page . . .

4.6.1.7 Inherited disorders

BL2HP

Question 7 continued

question	answers	extra information	mark
7(c)	**Argued evaluation** any **four** from: • PGD higher financial cost • PGD occurs before pregnancy / implantation • PGD does not involve abortion so less trauma / less pain / ethical comparison • PGD higher incidence of false positive / use of numbers so higher risk of destroying healthy embryo • PGD no chance of miscarriage whereas CVS does **or** PGD less chance of miscarriage	accept CVS only costs £600 accept detected at earlier stage so less unethical / less trauma accept PGD has (surplus) embryos so some destroyed / unethical	4
Total			**10**

UMS Conversion Calculator www.aqa.org.uk/umsconversion

4.6.1.7 Inherited disorders

Answer 07 · Mark scheme

Question 6

question	answers	extra information	mark
6(a)(i)	allele expressed even when other allele present **or** expressed if just one copy of allele is present **or** expressed if heterozygous	if present other allele not expressed	1
6(a)(ii)	2 affected parents have unaffected child **or 1** and **2** → **5** / **6** **or** if recessive all of **1** and **2**'s children would have CADASIL		1
6(a)(iii)	heterozygous – has unaffected children **or** because if homozygous all children would have CADASIL		1
6(b)	genetic diagram including: correct gametes: **D** and **d** **and** **d** (and **d**) derivation of offspring genotypes: **Dd** **Dd** **dd** **dd** identification of **Dd** as CADASIL **or dd** as unaffected correct probability: 0.5 / ½ / 1 in 2 / 50 % / 1 : 1	accept alternative symbols, if defined ignore 7 / 8 or male / female allow just **Dd** **dd** if ½-diagram allow ecf if correct for student's gametes allow ecf if correct for student's gametes	1 1 1 1
6(c)(i)	stem cells can differentiate **or** are undifferentiated / unspecialised can form blood vessel cells / brain cells **or** stem cells can divide		1 1

Question 6 continues on the next page . . .

4.6.1.7 Inherited disorders

BL2HP

Question 6 continued . . .

question	answers	extra information	mark
6(c)(ii)	ethical argument – eg no risk of damage to embryo **or** adult can give consent for removal of cells **or** adult can re-grow skin **or** if from a relative then less chance of rejection **or** if from self then no chance of rejection **or** skin cells more accessible	more ethical qualified ignore religion unqualified	1
Total			**10**

4.6.1.7 Inherited disorders

Answer 07 · Mark scheme

January 2013 | BL2HP | Original Q6 | Legacy

BL2HP

4.6.1.7 Inherited disorders

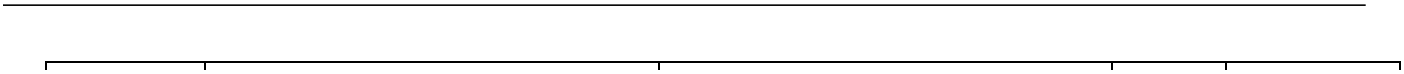
Answer 08 · Mark scheme June 2015 | BL2HP | Original Q3 | Legacy

Question	Answers	Extra information	Mark	AO / Spec. Ref.
3(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
3(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
3(b)(i)	1 / one		1	AO2 2.7.2c,e, 2.7.3a,c
3(b)(ii)	2 / two		1	AO2 2.7.2c,d, 2.7.3a,b
3(c)	**B**		1	AO3 2.7.2i
Total			**6**	

4.6.1.7 Inherited disorders

Answer 08 · Mark scheme

June 2015 | BL2HP | Original Q3 | Legacy



4.6.1.7 Inherited disorders

Answer 09 · Mark scheme

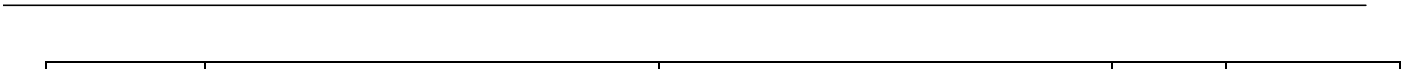
June 2016 | BL2HP | Original Q6 | Legacy

Question	Answers	Extra information	Mark	AO / Spec. Ref.
6(a)(i)	man has (inherited) polydactyly (PD) allele (from mother) man has (inherited) other / normal / recessive allele from father because father does not have PD allele **or** if father had it father would have had PD **or** father only has normal allele **or** father is homozygous recessive	allow gene for allele	1 1 1	AO1/2/3 2.7, 2.7.2a/c/d 2.7.3a/b
6(a)(ii)	0.5 / ½ / 1 in 2 / 1:1 / 50%	do not allow 1:2 or 50/50 allow 50:50	1	AO3 2.7, 2.7.2a/c/d 2.7.3a/b
6(b)	parental phenotypes: both brown parental genotypes: both **Bb** gametes: **B** **b** and **B** **b** offspring genotypes: **BB** **(2)Bb** **bb** offspring phenotypes correctly assigned to genotypes: **BB** & **Bb** = brown **bb** = red	allow only on gametes answer line allow ecf from genotypes allow ecf from gametes do not penalise confusion of 'phenotypes' & 'genotypes' here	1 1 1 1 1	AO1/2/3 2.7, 2.7.2a/c/e
Total			**9**	

4.6.1.7 Inherited disorders

Answer 09 · Mark scheme

June 2016 | BL2HP | Original Q6 | Legacy



4.6.1.7 Inherited disorders

Answer 10 · Mark scheme

Question	Answers	Extra information	Mark	AO / Spec. Ref.
06.1	DNA / deoxyribo(se)nucleic acid		1	AO1 4.1.2.1
06.2	controls the movement of substances in / out of cell	ignore allows substances in / out of cell	1	AO1 4.1.1.2
06.3	**Level 3:** Relevant points (reasons / causes) are identified, given in detail and logically linked to form a clear account.		5–6	AO2 4.2.2.1 4.2.2.2 4.4.2.1
	Level 2: Relevant points (reasons / causes) are identified, and there are attempts at logical linking. The resulting account is not fully clear.		3–4	
	Level 1: Points are identified and stated simply, but their relevance is not clear and there is no attempt at logical linking.		1–2	
	No relevant content		0	
	Indicative content fewer (digestive) enzymes enter small intestine • (so) enzymes break down / digest less food • (therefore) less absorption of nutrients • (so) less glucose can enter the blood(stream) • (so) less (glucose) available for respiration • (less absorption of nutrients so) fewer amino acids can enter the blood(stream) • (so) less protein is made (for growth) ignore ref to bile less oxygen enters blood(stream) / reaches cells / organs / tissues (ignore body / lungs) • (so) less (oxygen) available for (aerobic) respiration • (so) less energy released / available (for growth) do **not** accept energy produced for **level 3**, reference to both enzymes and oxygen is required			

4.6.1.7 Inherited disorders

Answer 10 · Mark scheme

June 2018 | 8461/1H | Original Q6

Question	Answers	Extra information	Mark	AO / Spec. Ref.
06.4	put ages in (ascending / descending / chronological) order		1	AO2 4.2.2.5
	find middle value	allow a way of achieving this ie $\frac{(n+1)}{2}$ where n is the number of people	1	AO1 4.2.2.5
06.5	most common in young people **or** (more) don't live to an old age	allow causes of this	1	AO3 4.2.2.5
06.6		an answer of 10 584 scores **2** marks		AO2 4.2.2.5
	$0.98 \times 10\,800$	allow $\frac{98}{100} \times 10\,800$	1	
	10 584	allow $10\,800 - (0.02 \times 10\,800)$ or equivalent	1	
06.7	take stem cells from patient (bone marrow / skin)		1	AO2 4.1.2.3
	remove / change / fix the faulty / CF gene		1	
	create embryo using these stem cells	allow idea of transferring nucleus from stem cells into enucleated egg / ovum	1	
	remove stem cells (from embryo) and stimulate / allow them to divide / differentiate into lung (cells)	if no other marks awarded allow **1** mark for therapeutic cloning	1	

4.6.1.7 Inherited disorders

Answer 10 · Mark scheme

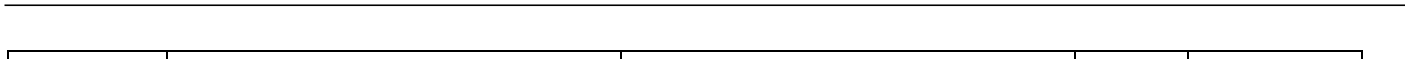
June 2018 | 8461/1H | Original Q6

Question	Answers	Extra information	Mark	AO / Spec. Ref.
06.8	idea of ethical / religious concerns about use of embryos such as loss of (potential) human life	allow idea that embryo cannot give consent ignore idea of against human rights of embryo unqualified ignore religious / ethical reasons unqualified	1	AO3 4.1.2.3
Total			18	

4.6.1.7 Inherited disorders

Answer 10 · Mark scheme

June 2018 | 8461/1H | Original Q6



4.6.1.7 Inherited disorders

Answer 11 · Mark scheme

November 2021 | 8461/2H | Original Q8

Question	Answers	Extra information	Mark	AO / Spec. Ref.
08.1	mutation means less oxygen for (aerobic) respiration	allow haemoglobin **or** red blood cell carries oxygen for (aerobic) respiration do **not** accept no oxygen for respiration	1	AO2 4.6 4.6.1.5 4.2.2.3
08.2	4 ÷ 17 0.235(29...)	allow 4:13 allow 0.24 **or** 24% allow ratio 1 : 3.25	1 1	AO2 4.6.1.5 4.2.2.3
08.3	father / **8**'s gametes correct: $H^A + H^A$ mother / **9**'s gametes correct: $H^A + H^S$	allow **1** mark for both sets of gametes if parents not identified	1 1	AO2 AO2
	correct derivation of offspring genotypes: H^AH^A H^AH^A H^AH^S H^AH^S	allow correctly derived offspring genotypes from incorrect parental gametes	1	AO2
	correct phenotype for each derived genotype		1	AO2
	0.25 / ¼ / 25% / 1 in 4 / 1:3	allow **only** a probability consistent with student's derivations	1	AO3 4.6.1.6 4.6.1.7 4.6.1.8

4.6.1.7 Inherited disorders

Answer 11 · Mark scheme

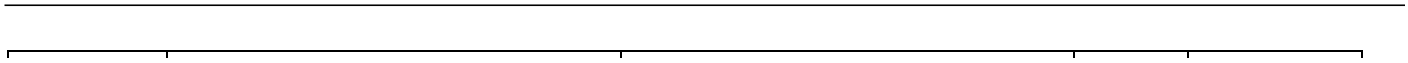
November 2021 | 8461/2H | Original Q8

Question	Answers	Extra information	Mark	AO / Spec. Ref.
08.4	any **three** from: *points for:* • H^AH^S do not get malaria • H^AH^S survive sickle cell anaemia *points against:* • H^AH^A may die from malaria • H^SH^S may become (severely) ill with sickle cell anaemia • Judgement: if parents H^AH^S then some offspring survive both malaria and sickle cell anaemia **or** if parents H^AH^S then some offspring may become (severely) ill with malaria and some become (severely) ill with sickle cell anaemia	to gain full marks both point(s) for and point(s) against must be given	3	AO3 4.6.2.1
Total			**11**	

4.6.1.7 Inherited disorders

Answer 11 · Mark scheme

November 2021 | 8461/2H | Original Q8



4.6.1.7 Inherited disorders

Answer 12 · Mark scheme

June 2024 | 8461/1H | Original Q2

Question 2

Question	Answers	Extra information	Mark	AO / Spec Ref.
02.1	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.
02.2	**A**, **D** and **E**		1	AO1 4.2.2.1 4.2.2.2

4.6.1.7 Inherited disorders

Answer 12 · Mark scheme

Question	Answers	Mark	AO / Spec Ref.
02.3	**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 12 · Mark scheme

Question	Answers	Extra information	Mark	AO / Spec Ref.
02.4	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 12 · Mark scheme

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

Total Question 2	14

4.6.1.7 Inherited disorders

Answer 13 · Mark scheme

June 2024 | 8461/2H | Original Q6

Question 6

Question	Answers	Extra information	Mark	AO / Spec Ref.
06.1	(an allele) that is always expressed **or** (an allele) that is expressed even when the other / recessive allele is present **or** (an allele) that is expressed in the heterozygote **or** (an allele) that is expressed when only one copy is present	allow always shows in the phenotype ignore stronger	1	AO1 4.6.1.6

Question	Answers	Extra information	Mark	AO / Spec Ref.
06.2		allow an annotated genetic diagram for up to **2** marks		AO3 4.6.1.6 4.6.1.7
	(person **1**) has polydactyly so must have **D**	allow (person **1**) has polydactyly so must have a dominant allele	1	
	has offspring / (person) **5** who does not have polydactyly so must have **d** from person **1** **or** person **5** does not have polydactyly so must be **dd** and must inherit **d** from person **1**		1	

4.6.1.7 Inherited disorders

Answer 13 · Mark scheme

Question	Answers	Extra information	Mark	AO / Spec Ref.
06.3	Female / **6**'s gametes correct **D + d** Male / **7**'s gametes correct **d + d**	allow **1** mark for both sets of gametes if parents not identified	1 1	AO2 4.6.1.6 4.6.1.7
	correct derivation of offspring genotypes: **Dd Dd dd dd**	derivation must be consistent with parental gametes	1	
	Dd correctly identified as polydactyly in **only** half of offspring	mp4 only awarded if mp3 is correct	1	

Question	Answers	Extra information	Mark	AO / Spec Ref.
06.4	CF allele is recessive	allow CF is recessive	1	AO1
	to have CF, must have 2 CF alleles		1	AO2
	chance of having one CF allele is $\frac{1}{50}$	ignore chance of having one CF allele is one in 50	1	AO2
	(chance of having two CF alleles is) $\frac{1}{50} \times \frac{1}{50} = \frac{1}{2500}$	ignore 50 × 50 = 2500	1	AO2 4.6.1.6 4.6.1.7

Total Question 6	11

4.6.1.7 Inherited disorders

Answer 10 · Examiner comments

June 2018 | 8461/1H | Original Q6

Question 6 (standard, standard / high & high demand)

Following updated research information and communications with parents of children with cystic fibrosis, we have decided to withdraw the question on this topic from this paper.

It has been decided not to provide a replacement question as it would not be possible to determine accurately how a replacement question would have performed if it had been part of the original paper taken in 2018.

The following statistics may help you to make effective use of the remainder of the paper:

Mean mark (maximum mark)	
06.1	0.47 (1)
06.2	0.65 (1)
06.3	2.27 (6)
06.4	1.27 (2)
06.5	0.69 (1)
06.6	1.33 (2)
06.7	0.56 (4)
06.8	0.42 (1)
Question 6	7.66 (18)

4.6.1.7 Inherited disorders

Answer 11 · Examiner comments

Question 8 (standard/high & high demand)

This question was about the inheritance of sickle cell anaemia and its effects. Answers to **08.1** were generally insufficient. Many students knew that a mutation in the haemoglobin gene could result in less oxygen being transported around the body (although some stated this would 'affect' the amount of oxygen), but hardly any went on to explain that this would mean a lower level of respiration.

Calculations in **08.2** were weak. Some students did not count the number of normal and sickled cells in Figure 13. Many did not understand the difference between the ratio 4:13 and the fraction $\frac{4}{13}$. Many did not go on to convert the correct fraction $\frac{4}{17}$ to a decimal (0.24) or to a percentage (24%). Some students, having worked out a percentage, did not include the % symbol in their answer, thus giving an incorrect proportion of '24'. Thus few students scored both marks.

08.3 was a construction of a Punnett square to determine the probability that certain parents from the family pedigree diagram would produce a child with sickle cell trait, followed by multiplication by ½ to find the probability that this child would be female. A very small proportion of students was completely successful. Errors included assigning the same genotype to parents who had different phenotypes, not stating which gametes came from which parent, not identifying the phenotypes of the offspring genotypes and not dividing the probability from the Punnett square by 2 to allow for the biological sex specified in the question. A few students showed the offspring as being haploid even though each was derived from two haploid gametes.

In **08.4**, students had difficulty in applying information from various other parts of question 8 to evaluate the scientist's statement about the advantage of having the H^S allele in countries where malaria occurs. Around 96% of studebts did not discuss the effects of sickle cell anaemia and malaria on people of different stated genotype and thus scored no marks.

4.6.1.7 Inherited disorders

Answer 12 · Examiner comments

Question 2 (standard demand)

02.1 Most of the students who gained this mark (93%), identified 'nucleus', with many also suggesting 'chromosome'. 'DNA' formed part of the response for a significant proportion of students, but this was often associated with either of the two acceptable answers, so credit could be awarded. Very few students gave alternative components of a cell.

02.2 98% of students identified organs A, D and E as being the lungs, small intestine and pancreas.

02.3 This extended response question provided students with the opportunity to express their knowledge of the enzymes produced by the pancreas and how the products of digestion are used within the body. However, as this question was phrased in the context of a person with cystic fibrosis, marks could only be gained by students who described the details they had revised not occurring. Thus, those who only described what normally goes on in the body without any indication that in this context, these processes would not occur or would be reduced, could not gain credit. Students went about describing their answers in two ways: either giving the details they knew, followed by a statement that these processes would not happen or happen less, or by describing each step in terms of reduction (eg 'less amylase is produced so less starch is digested into glucose').

Lower-attaining students often got no further than repeating the details they had been given; 'less of each enzyme, so less digestion occurs', was a common response by many students, although they often repeated this in multiple ways to fill up the available space. It was also noticeable that lower-attaining students got no further than repeating the information in second bullet point from the question and did not attempt to respond to the command word of the question, 'explain'.

Although knowledge of the enzymes and their actions was generally good, the impact their lack would have on the body's ability to gain mass was less well understood. Few students referred to respiration, energy or the reasons why amino acids are needed by the body or cells. However, many did describe the reduction of absorption, often including the products of digestion in this story, and so could gain access to Level 3 as they had answered both parts of the question. Many students' answers lacked precision, describing the lack of absorption of starch, fat or protein, rather than of the products of the digestion of these large molecules, whilst others went on the describe the egestion of non-digested molecules; neither of which idea was creditworthy.

© 2024 AQA and its licensors. All rights reserved.

Those who only gave an answer to one side of the question (almost always the difficulty digesting food), however detailed this was, could not score higher than a mark in Level 2. Hence, students should be reminded, once they have completed their responses to questions such as this, to re-read the question, to ensure they have answered all parts of it.

Less than one third of students scored full marks in this question, with 13% scoring zero.

02.4 This question was generally well answered with nearly 50% of students gaining at least two marks. Despite the command word, 'describe', many students also added explanatory detail, often regarding diffusion in some way, but as the question did not ask for explanations, these were ignored. A 'large surface area' and a 'good blood supply' were frequently given. However, having 'thin walls' or having 'walls only one cell thick' was often poorly expressed, with answers implying that the alveoli themselves were thin or that they have a thin membrane; neither of which are correct. A few students drifted into an insecure knowledge of plant cell structure, describing the presence of 'thin cell walls', which was also not creditworthy.

It is a long-held and often repeated misconception that the alveoli being moist aids gas exchange. This is not correct and did not gain credit, although if included in an answer with three creditworthy points, was ignored.

02.5 Students could tackle this question in either of two routes: the reduction in (aerobic) respiration and its consequences or the occurrence of anaerobic respiration and its consequences. For many students, their responses contained elements of both routes, in which case the description which gave the higher mark was credited.

It should be noted that a reduction of oxygen supply will not stop (aerobic) respiration occurring altogether, so students who described this idea could not gain credit for this first point, although they might still have scored full marks for descriptions involving anaerobic respiration. In describing either route, 'tiredness' and 'fatigue' were common suggestions, however, without further qualification in terms of 'muscle' in the anaerobic respiration route, could not gain credit. Lower-attaining students gave descriptions which contradict the principle of conservation of energy, suggesting that energy is produced, rather than being transferred or released. Others suggest that energy is used *for* respiration, rather than being released by respiration.

27% of students scored full marks in this question, 35% scored two marks, and 22% one mark.

© 2024 AQA and its licensors. All rights reserved.

4.6.1.7 Inherited disorders

Answer 13 · Examiner comments

Question 6 (standard, standard/high and high demand)

06.1 Almost 45% of students were able to give a creditworthy answer having clearly learned a suitable definition, although the term 'expressed' was used less frequently by students than other synonyms. Numerous students remain confused between a gene and an allele. References to 'stronger/overpowering' alleles or suggesting they were 'always inherited passed on' did not gain credit.

06.2 Students were asked to explain how the inheritance chart provided shows that person 1 is heterozygous. Many students found it difficult to articulate their responses, with only 30% of students gaining 1 or both marks. Many students did not answer the question directly, but instead explained why the parent could not be a genotype other than heterozygous, rather than showing that they are heterozygous and did have polydactyly. Some students showed a clear understanding of the inheritance of the condition, while many students only gave incomplete answers that involved only part of each marking point.

06.3 55% of students gained all four marks by drawing and fully annotating a Punnett square to explain how the doctor determined a probability of 0.5. The 25% of students that scored three marks commonly did so for either not fully annotating their Punnett square identifying the genotypes of person 6 and 7 explicitly, or they did not identify which offspring were polydactyly. A minority of students did not gain marks by choosing to not use the letters 'D' and 'd' for their alleles, but instead chose a different letter without providing a key.

06.4 Many students correctly identified CF as a recessive condition or allele for marking point 1, and many went on to get marking point 2 also by stating that two copies of the allele are needed. Students that used the biological term 'homozygous recessive' in their explanation were awarded both marking points.

Very few students were awarded marking point 3 without marking point 4. There was clear confusion again here between the terms 'allele' and 'gene', despite the information in the question stem. A number of students gained marking points 3 and 4 with no reference to marking point 1 or 2. 15% of students scored the full four marks available in this question.

© 2024 AQA and its licensors. All rights reserved.