Question Number	Answer	Additional guidance	Mark
1(a)	1. (the disorder results from a) defect in genes / eq ; 2. both (defective) alleles need to be present / homozygous / not expressed in the presence of a dominant allele / eq ;	1. AL W faulty allele	**(2)**

Question Number	Answer	Mark
1(b)(i)	A ;	**(1)**

Question Number	Answer	Mark
1(b)(ii)	C ;	**(1)**

Question Number	Answer	Mark
1(b)(iii)	A ;	**(1)**

Question Number	Answer	Mark
1(b)(iv)	D ;	**(1)**

Question Number	Answer	Additional guidance	Mark
1(c)QWC	(QWC– Spelling of technical terms must be correct and the answer must be organised in a logical sequence) 1. {isolation / identification / eq} of normal gene / eq ; 2. {inserted / eq} into vector / stem cells / eq ; 3. vector named as {liposome / virus} ; 4. injection of {vector / modified stem cells} into {blood / brain / target cells / eq} / eq ; 5. ref to use of control injection ; 6. further detail of control injection e.g. use empty liposome / virus without gene inserted ; 7. progression of disease monitored / eq ; 8. life spans recorded / eq ; 9. reference to appropriate comparison with control eg untreated sheep ; 10. idea that treatment needs to be repeated; 11. idea of replication of investigation;	**QWC** penalise once if mark point is not in a logical position	**(5)**

Question Number	Answer	Mark
2(a)(i)	1. reference to alteration in DNA ; 2. change in {base (sequence) / quantity of DNA} / eq ;	**(2)**

Question Number	Answer	Mark
2(a)(ii)	idea that both of these alleles need to be present in order for the recessive phenotype to be expressed ;	**(1)**

Question Number	Answer	Mark
2(b)	1. idea of a gene being a sequence of bases that code for the sequence of amino acids in the {protein / polypeptide chain /enzyme / galactocerebrosidase} ; 2. (gene) mutation will alter {DNA triplet / DNA code / codon / eq} / eq ; 3. this may result in a different {amino acid / stop codon / amino acid sequence / primary structure / eq} / eq ; 4. idea that this may change the {shape / eq} of {protein / enzyme} ; 5. therefore causing {no synthesis / incomplete / eq} of {enzyme / galactocerebrosidase} / change of active site / eq ;	**(3)**

Question Number	Answer	Mark
2(c)	1. genotype of parents shown ; 2. alleles in the gametes shown ; 3. possible genotypes of children shown ; 4. corresponding phenotypes shown ; 5. (probability =) ¼ / 25% / 1 in 4 / 0.25 ;	**(5)**

Question Number	Answer	Mark
2(d)	amniocentesis / chorionic villus sampling / CVS / eq ;	**(1)**

Question number	Answer	Mark
3(a)(i)	1. idea that people are more likely to have obsessive compulsive disorder if they have a close relative with the condition ; 2. credit manipulation of figures i.e. 100 times more likely ; 3. therefore they may also have the {genes / alleles / genotype / eq} for this condition / eq ;	(2)

Question number	Answer	Mark
3(a)(ii)	1. idea that if they have a close relative with this illness, the risk {ought to be higher than / is only} 10% ; 2. therefore other factors must be involved as well / eq ; 3. named example of environmental influence e.g. learnt behaviour ;	(2)

Question number	Answer	Mark
3(a)(iii)	1. neurotic depression ; 2. {little / eq} difference between {population as a whole / females} and close relative data / eq ;	(2)

Question Number	Answer	Mark
***4(a) QW**	(QWC – Spelling of technical terms *(shown in italics)* must be correct and the answer must be organised in a logical sequence) 1. idea of (*mutation* / named mutation) causing different base sequence ; 2. reference to different {sequence of *amino acids* / *primary* structure} / eq ; 3. reference to {β chain / *haemoglobin* / *protein* / *polypeptide*} being the wrong shape ; 4. *haemoglobin* no longer binds oxygen / binds less *oxygen* / eq ; 5. {less / no } *oxygen* {supplied / carried / eq} (to the cells) / eq ; 6. correct reference to *respiration* / eq ; 7. idea of breathlessness due to body trying to take in more *oxygen* ; 8. idea of tiredness due to lack of energy ;	**max (4)**

Question Number	Answer	Mark
4(b)	<table><tr><td>25(%)</td><td>25(%)</td><td>50(%)</td></tr><tr><td>no chance / 0 (%)</td><td>no chance / 0 (%)</td><td>100 (%)</td></tr></table> All 3 in a row = 2 marks 1 or 2 in a row correct = 1 mark	**(4)**

Question Number	Answer	Mark
4(c) QWC	1. reference to use of {normal / correct} {allele / gene}; 2. for {haemoglobin / ß chain}; 3. reference to introduction of {gene / allele/ DNA} into cells ; 4. cells named as (bone) marrow / eq ; 5. reference to use of vector (to introduce gene into cells) ; 6. (named vector) e.g. virus, liposome ; 7. credit reference to appropriate mode of delivery of vector e.g. injection into (bone) marrow ; 8. reference to need for repeated treatment ;	**max (4)**