

Question Number	Answer	Additional Guidance	Mark
1(a)(i)	<ol style="list-style-type: none"> <li>idea of it being frequently inherited in the family e.g. 1, 7 and 10 all have affected offspring, 9 of the family have the disorder ;</li> <li>individual { 1 / 3 / 7 / 10 } must be heterozygous / eq ;</li> <li>use of the pedigree diagram to explain mark point 2 ;</li> <li>appropriate use recessive allele argument e.g. if it were recessive then 2 would have to be {heterozygous / a carrier} ;</li> <li>idea that it is unlikely that the unrelated parents { 8 / 11 } would also be carriers of the affected allele ;</li> </ol>	<p><b>IGNORE</b> 50% are affected</p> <p><b>1 ACCEPT</b> 8 of 14 descendants of 1 and 2</p> <p><b>2 ACCEPT</b> have one dominant allele</p>	(3)

Question Number	Answer	Additional Guidance	Mark
1(a)(ii)	<ol style="list-style-type: none"> <li>parents gametes displayed correctly ( e.g. M, m and m ,m) ;</li> <li>correct genotypes of offspring shown ;</li> <li>probability matches genotypes shown e.g. 0.5 / ½ / 1 in 2 / 50% ;</li> </ol>	<p><b>1. ACC T</b> gametes shown on a punnett square alone</p> <p><b>3. ACCEPT</b> other probabilities if match genotypes shown</p>	(3)

Question Number	Answer	Additional Guidance	Mark
1(b)	<p>Any two pairs from:</p> <ol style="list-style-type: none"> <li>1. idea that there is a { thick wall / thick layers / thick tunica media / eq } ;</li> <li>2. idea that it needs to { avoid rupture / withstand high pressure / allow expansion / eq } ;</li> <li>3. collagen / elastic fibres ;</li> <li>4. allow expansion / elastic recoil ;</li> <li>5. muscle { layer / fibres / wall / eq } ;</li> <li>6. control the flow of blood / maintain blood pressure / eq ;</li> <li>7. smooth endothelial wall / eq ;</li> <li>8. to reduce { friction / resistance / eq } ;</li> <li>9. narrow lumen ;</li> <li>10.(to maintain) high blood pressure / eq ;</li> </ol>	<p><b>3 and 4 . ACCEPT</b> folded {endothelium / (inner) surface} allows expansion</p>	(4)

Number			
<b>2(a)</b>	<ol style="list-style-type: none"> <li>1. idea of more than one gene for a single { characteristic / trait } ;</li> <li>2. on more than one locus ;</li> <li>3. idea of continuous variation ;</li> <li>4. idea of genes interacting with each other ;</li> </ol>	IGNORE phenotype	<b>(2)</b>

Question Number	Answer	Additional Guidance	Mark
<b>2(b)(i)</b>	a Himalayan rabbit shaved (in the same place) and no ice pack (taped to bald patch);	<b>ACCEPT</b> shaved with no ice or another object taped to its back instead of an icepack	<b>(1)</b>

Question Number	Answer	Additional Guidance	Mark
<b>2(b)(ii)</b>	<ol style="list-style-type: none"> <li>1. fur grew black when exposed to cold temperatures / eq ;</li> <li>2. fur remains white when not exposed to cold temperatures / eq ;</li> <li>3. idea that the gene is { expressed / activated } at low temperatures ;</li> </ol>	2. ACCE fur is white in warm areas	<b>(3)</b>

Question Number	Answer	Additional Guidance	Mark
<b>2(c)</b>	<ol style="list-style-type: none"> <li>1. fur is (only) black where { the temperature is lower than 25 °C / ice pack is placed } ;</li> <li>2. because the enzyme is active / eq ;</li> </ol>	1. ACCE darker fur	<b>(2)</b>

Number			
<b>3(a)</b>	<ol style="list-style-type: none"> <li>{ identical / monozygotic twins } are genetically identical / eq ;</li> <li>derived from one egg and one sperm /one { zygote / embryo / eq } / eq ;</li> <li>(so any phenotypic ) difference is due to { nurture / environmental } / eq ;</li> <li>{ non-identical twins / dizygotic twins} are genetically different ;</li> <li>(any phenotype) that is different when the environment is the same is likely to be { nature / genetic / eq } / eq ;</li> </ol>	<p><b>ACCEPT</b> comments on monozygotic twins (MZ) raised apart as a context</p> <p><b>1 ACCEPT</b> same alleles <b>IGNORE:</b> same DNA / genes</p> <p><b>2 ACCEPT</b> one fertilised egg, ball of cells, blastula</p>	<b>(4)</b>

Question Number	Answer	Additional Guidance	Mark
<b>3(b)(i)</b>	<ol style="list-style-type: none"> <li>study groups from different cultures / eq ;</li> <li>(if) outcome is the same then (likely to be) nature ;</li> <li>(if) outcome is different in the groups then (likely to be) nurture ;</li> </ol>	<b>1 IGNORE</b> different countries / environments	<b>(2)</b>

Question Number	Answer	Additional Guidance	Mark
<b>3(b)(ii)</b>	<ol style="list-style-type: none"> <li>idea of large sample size ;</li> <li>idea of standardised sampling technique e.g. age, gender ;</li> <li>same (range of) emotions used / eq</li> </ol> <p>OR</p> <p>standard methodology e.g. same photos ;</p>		<b>(2)</b>

Question Number	Answer	Additional Guidance	Mark
4(a)	1. idea that both of these alleles need to be present in order for the recessive phenotype to be expressed ;  <b>AND any two of:</b>  2. different form of a gene / eq ; 3. same locus /position / eq ; 4. different base sequence / eq ;	1. ACCEPT not expressed in presence of dominant allele  2. ACCEPT type of same gene NOT just type of gene	(3)

Question Number	Answer	Additional Guidance	Mark
4(b) (i)	1. genotypes and phenotypes of parents ;  2. gametes clearly shown as individual alleles ;  3. offspring genotypes ;  4. phenotypes correctly matched to genotypes ;	IGNORE gender  1. & 4. ACCEPT carrier/ normal / healthy / unaffected /sufferer as a phenotype  2. OT E.C.F. from 1. 2. & 3 can be awarded in a Punnett square  3. and 4. can be awarded as E.C.F. from 2	(4)

Question Number	Answer	Additional Guidance	Mark
4(b) (ii)	0.5 / ½ / 1 in 2 / 50% ;	IGNORE expressed as a ratio	(1)

Question Number	Answer	Additional Guidance	Mark
5(a)	1. homozygous ; 2. channel / transport / transmembrane / intrinsic / globular ; 3. chloride / $\text{Cl}^-$ / Cl ; 4. reproductive / eq ;	1. <b>ACCEPT</b> (a) homozygote  3. <b>DO NOT ACCEPT</b> chlorine	(4)

Question Number	Answer	Additional Guidance	Mark
5(b)	1. produces { thicker / stickier / more viscous / eq } mucus ; 2. blocking { trachea / bronchi / bronchioles / airway / eq } / eq ; 3. cilia are unable to move mucus out of lungs / eq ; 4. idea of reduced flow of { air / oxygen } to alveoli ; 5. idea of reduced concentration gradient for { oxygen / carbon dioxide } (in alveoli) ; 6. idea of loss of surface area / elasticity / eq ; 7. idea of reduced gaseous exchange ; 8. trapped bacteria may result in more respiratory infections / eq ;	1. <b>CCEPT</b> sticky / thick in context, <b>ACCEPT</b> less water in mucus 2. <b>IGNORE</b> respiratory system <b>ACCEPT</b> alveoli  7. <b>CCEPT</b> less $\text{O}_2$ diffuses into blood <b>IGNORE</b> larger diffusion pathway	(4)

Question Number	Answer	Additional Guidance	Mark
5(c) (i)	1. chorionic villus sampling / amniocentesis ; 2. idea that (fetal) {cells / DNA} are obtained from appropriate source { placenta / amniotic fluid / eq } ; 3. (cells / DNA) tested for presence of {CFTR / recessive / faulty / mutant / eq} {allele / gene } / eq ;	1. <b>ACCEPT</b> CVS <b>DO NOT ACCEPT</b> chronic 2. <b>ACCEPT</b> from embryo 3. <b>ACCEPT</b> test for cystic fibrosis allele or gene	(3)

Question Number	Answer	Additional Guidance	Mark
5(c) (ii)	Any of the following paired points 1. idea that it may result in a miscarriage / choice of an abortion ; 2. {killing / eq} is {wrong / unethical / eq}; <b>OR</b> 3. idea of risk of false {positive / negative} ; 4. comment on consequence e.g. healthy fetus may be aborted / parents not prepared for child with cystic fibrosis / eq ; <b>OR</b> 5. if cystic fibrosis or some other abnormality may be found ; 6. comment on possible problems with {future employment / insurance / what constitutes a serious condition} / eq ; <b>OR</b> 7. who has right to decide if tests should be performed / eq ; 8. {implications of medical costs / disagreements over next step} ; <b>OR</b> 9. issues relating to confidentiality of {parents / child} / eq ; 10. idea that {some other abnormality may be found / paternal DNA does not match / other family members have right to know results} ;	1. <b>ACCEPT</b> can {harm / damage / kill} the fetus 2. <b>ACCEPT</b> fetus has right to life / distress to parents / genetic discrimination / eugenics 3. <b>ACCEPT</b> it isn't 100% accurate 4. <b>ACCEPT</b> parents did not have choice of abortion	(2)