

**Assessment Schedule – 2022****Science: Demonstrate understanding of biological ideas relating to genetic variation (90948)****Evidence Statement**

	<b>Evidence</b>	<b>Achievement</b>	<b>Merit</b>	<b>Excellence</b>
ONE (a)	A mutation is a change in the order of DNA bases which causes a new version of the gene called an allele. The sickle cell disease mutation allele codes for the phenotype “sickle cell disease”.	<ul style="list-style-type: none"> <li>Defines mutation as a change in the DNA / gene / base sequence. <i>(May come from a or b)</i></li> </ul>	<ul style="list-style-type: none"> <li>A mutation is a change in the order of DNA bases / gene which codes for a new allele.</li> </ul>	<ul style="list-style-type: none"> <li>A mutation causes a change in the order of DNA bases in the gene coding, which may result in a new allele giving the phenotype which is sickle cell disease.</li> </ul>
(b)	A gene is a section of DNA that codes for unaffected or affected sickle cell disease trait. A mutation is a change in the order of DNA bases which causes a new version of the gene called an allele. The new allele codes for the phenotype “sickle cell disease”. DNA carries genetic information as a base code.	<ul style="list-style-type: none"> <li>Defines DNA as genetic material.</li> <li>Gene as a section of DNA that codes for a trait / protein / phenotype.</li> <li>Allele is a different form of a gene.</li> <li>Define phenotype (physical expression of genotype / gene / sickle cell disease).</li> </ul>	<ul style="list-style-type: none"> <li>If the unaffected (normal) gene is changed / mutated it may create a new sickle cell / phenotype. <i>M1 and 2 may be found in part a.</i></li> </ul>	<ul style="list-style-type: none"> <li><i>(must link phenotype to sickle cell)</i></li> </ul>
(c)	If a mutation coding unaffected / affected by Sickle cell disease occurs in the gametes it can be passed down in the DNA from either parent when sperm and egg (gametes) fuse during fertilisation.	<ul style="list-style-type: none"> <li>Described as passing on in the sperm / egg / genes.</li> </ul>	<ul style="list-style-type: none"> <li>A mutation can only be passed down if it occurs in the gametes / not in somatic cells. <i>(not sperm and egg)</i></li> <li>Can be passed down in the DNA when sperm and egg (gametes) fuse / during fertilisation.</li> </ul>	<ul style="list-style-type: none"> <li>A mutation must occur in the gametes for it to be passed down through fertilisation.</li> </ul>

<b>N0</b>	<b>N1</b>	<b>N2</b>	<b>A3</b>	<b>A4</b>	<b>M5</b>	<b>M6</b>	<b>E7</b>	<b>E8</b>
No response, or no relevant evidence.	ONE Achievement point.	TWO Achievement points.	THREE Achievement points.	FOUR Achievement points.	TWO Merit point.	THREE Merit points.	TWO Excellence points with minor omission. <i>(fertilisation)</i>	TWO Excellence points.

	Evidence	Achievement	Merit	Excellence																
TWO (a)	1 Hh (heterozygous) 2 Hh (heterozygous) 3 hh (homozygous recessive)	• Two genotypes correct.																		
(b)	<table><tr><td></td><td colspan="3"></td></tr><tr><td></td><td></td><td>H</td><td>h</td></tr><tr><td>H</td><td>HH</td><td>Hh</td><td></td></tr><tr><td>h</td><td>Hh</td><td>hh</td><td></td></tr></table>							H	h	H	HH	Hh		h	Hh	hh		• Two correct genotypes in same row or same column.		
		H	h																	
H	HH	Hh																		
h	Hh	hh																		
(c)	<table><tr><td></td><td>exp</td><td>actual</td></tr><tr><td>H:unaf fected</td><td>3:1</td><td>2:2 Or 1:1</td></tr></table>		exp	actual	H:unaf fected	3:1	2:2 Or 1:1	• Any ratio correct.												
	exp	actual																		
H:unaf fected	3:1	2:2 Or 1:1																		
(d)	<p>Expected ratio 3:1 Huntington’s to unaffected. Actual ratio 2:2 or 1:1 to Huntington’s : Unaffected respectively.</p> <p>I-1 and I-2 offspring have less Huntington’s disease than expected. This is because each offspring is the product of a random event / fertilisation.</p> <p>Each offspring is unaffected by previous outcomes. Since I-1 and I-2 are both heterozygous (Hh), each individual has 50% chance of inheriting either allele from each parent.</p> <p>With a larger number of offspring, we would expect a ratio very close to 3:1 to Huntington’s to unaffected respectively.</p>	• Describes random fertilisation. • Punnett square only shows possible / predicted outcomes. • Mentions small sample size.	• Explains actual ratio of Huntington’s is different than expected because each offspring / fertilisation depends on chance (random / unaffected by previous outcomes all OK). • Each (heterozygous) parent has a 50% chance of passing on either allele (or offspring have a 75% chance of inheriting H). • More offspring are needed to match the expected ratio	• Explains actual ratio of Huntington’s is different than expected because each offspring is the result of random fertilisation therefore has 50:50 chance of passing either allele and is unaffected by previous outcomes • Four offspring is a small sample, with a larger number of offspring we would expect a ratio very close to 3:1 / 75% by inheriting a dominant allele from either parent.																

(e)	<p>If Huntington's was recessive, two affected individuals I1 I2 would both have the genotype hh and hh. Thus they can pass only an allele h, and unaffected offspring can never be produced.</p> <p>OR</p> <p>Huntington's is a dominant disease because both I1 and I2 have the condition but pass on unaffected alleles to I3 or I6. This means they are heterozygous and the dominant allele H causes the condition.</p>	<ul style="list-style-type: none"> <li>States they have offspring which differs from themselves.</li> </ul> <p>OR</p> <p>The parents must have both alleles / Hh / heterozygous.</p>	<ul style="list-style-type: none"> <li>Explains that both parents must be heterozygous (Hh) as they are both affected by Huntington's and have unaffected children.</li> </ul> <p>OR</p> <p>If the parents were homozygous recessive (hh) then all of their children would be the same as them.</p>	<ul style="list-style-type: none"> <li>Links phenotypes, alleles passed on and gives examples from the pedigree chart.</li> </ul> <p><i>If parents were both hh (homozygous recessive) then the parents are pass on recessive alleles and all children would be affected (by Huntington's) and links to pedigree chart (provides examples)</i></p>
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NØ	N1	N2	A3	A4	M5	M6	E7	E8
No response, or no relevant evidence.	ONE Achievement point.	TWO Achievement points.	THREE Achievement points.	FOUR Achievement points.	ONE Merit points.	TWO Merit points.	ONE Excellence point.	TWO Excellence points

	Evidence	Achievement	Merit	Excellence
THREE (a)	<p>Process of fertilisation: Random male and female gametes with a unique half of the number of chromosomes from each parent fuse, producing a genetically unique zygote / offspring.</p> <p>Process of meiosis: Random assortment of chromosomes that line up along the middle of the cell before being separated. Crossing over of chromosomes to exchange genetic material in meiosis produce new combinations of alleles and increase genetic variation between individuals.</p>	<ul style="list-style-type: none"> <li>Defines random fertilisation.</li> <li>States gametes fuse during fertilisation.</li> <li>States gametes are genetically unique OR have half the chromosomes.</li> <li>Variation is a range of traits / phenotypes within a population (<i>May be accepted from part a or b.</i>)</li> </ul>	<ul style="list-style-type: none"> <li>Explains meiosis creates genetic variation by random assortment of DNA / crossing over / independent segregation chromosomes during meiosis.</li> <li>Explains <b>fertilisation</b> (egg and sperm fuse). and how its <b>random</b> nature creates <b>genetic variation</b>.</li> <li>Explains fertilisation and how <b>genes combine</b> from two parents / egg &amp; sperm create <b>genetic variation</b> (1/2 from each parent).</li> </ul>	<ul style="list-style-type: none"> <li>Discusses one way meiosis causes variation. AND Discusses how fertilisation cause genetic variation.</li> </ul>
(b)	<p><b>Genetic</b> variation in a population leads to varied phenotypes small / large beaks. This allows the population to survive in a changing / varied environment because large-beaked individuals are more likely to survive a drought because they can eat larger seeds and thus reproduce, passing on favourable combinations of large-beaked genes / alleles to the next generation. Whilst small-beaked birds find it difficult finding food and many die before passing on their genes.</p> <p>Over many droughts, large-beaked birds' genes / alleles will become more common in the population.</p>	<ul style="list-style-type: none"> <li>States birds with large beaks can survive by eating large (and small) seeds (or converse).</li> <li>Large-beaked birds reproduce more / passing on genes,</li> <li>Large-beaked birds will become more common if droughts continue (or converse).</li> </ul>	<ul style="list-style-type: none"> <li>Large-beaked birds more likely to <b>survive</b> (in a drought) as they are <b>better suited</b> to the environmental change (seeds).</li> <li>Large-beaked birds more likely to <b>survive</b> and reproduce / passing on their genes / alleles / trait (<i>not large beaks</i>).</li> </ul>	<ul style="list-style-type: none"> <li>Large-beaked birds more likely to <b>survive</b> (in a drought) as they are <b>better suited</b> to the environmental change (seeds) allowing them to <b>reproduce</b>, passing on the <b>allele / gene</b> (not trait) AND Over many years of droughts, large-beaked birds' will become more common.</li> </ul>

N0	N1	N2	A3	A4	M5	M6	E7	E8
No response, or no relevant evidence.	ONE Achievement point.	TWO Achievement points.	THREE Achievement points.	FOUR Achievement points.	TWO Merit points.	THREE Merit points.	ONE Excellence point.	TWO Excellence points.

### Cut Scores

Not Achieved	Achievement	Achievement with Merit	Achievement with Excellence
0 – 6	7 – 13	14 – 19	20 – 24