Assessment Schedule – 2025

Chemistry and Biology: Demonstrate understanding of genetic variation in relation to an identified characteristic (92022)

Assessment Criteria

Achievement	Achievement with Merit	Achievement with Excellence
Demonstrating understanding of genetic variation in relation to an identified characteristic involves: • describing the source and the nature of genetic variation using an identified characteristic • describing a purpose for identifying genetic relationships through the use of a gene tracking methodology.	*Explaining genetic variation in relation to an identified characteristic* involves: • explaining how and why the genetic variation occurs using an identified characteristic • explaining how the purpose for identifying genetic relationships through the use of a gene tracking methodology is met.	*Evaluating genetic variation in relation to an identified characteristic* involves: • evaluating findings when genetic variation has been identified and tracked for the purpose of identifying genetic relationships.

Cut Scores

Not Achieved	Achievement	Achievement with Merit	Achievement with Excellence
0–7	8–12	13–18	19–24

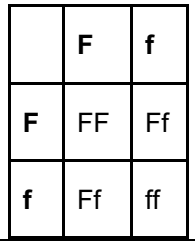
Evidence

Question One	Evidence	Achievement	Achievement with Merit	Achievement with Excellence
(a)	Chromosomes are long strands of DNA tightly wound together. DNA is a large molecule that is coiled up into a double-helix structure. It is made up of ribose / sugar, phosphate, and a base. There are four types of bases that always pair up (A with T and G with C). A gene is a specific section of the DNA sequence. It codes a specific trait. An allele is a different version of the same gene. Alleles provide some variation in the characteristic / phenotype. For example, human genomes have two copies of chromosome 7. These chromosomes are made up of tightly wound, long strands of DNA. The DNA contains bases that are in a particular order. A specific section of chromosome 7 contains the CFTR gene. The order of bases in this gene is the same for every copy that is a "normal allele, F". A slight change in these bases gives a different version of the gene – an allele. This is represented in Figure 1 as "mutant allele (f)".	Accurately defines OR describes: • chromosome • genes • alleles • DNA.	• Explains the relationship between chromosomes, genes, alleles, and DNA, using clear examples. *(Examples **do not** have to be within the CFTR gene context.)*	
(b)(i) (ii)	Mutation is a (permanent) base change in the sequence of a DNA strand. Genotype is the allele combination (for a specific trait) at a particular locus on a chromosome. Phenotype is the physical expression of the genotype – the observable characteristics or traits resulting from the interaction of that genotype with the environment. Genetic variation is the difference between the genes / alleles in a population. In this case, individuals can have one of three genotypes for the CFTR gene: FF, Ff, or ff. The FF and Ff genotypes will express a phenotype of normal mucus production, while individuals with the ff genotype will express a phenotype of excessive mucus production (cystic fibrosis). Genetic variation for the CFTR gene is observed in two different alleles. The normal allele (F) and the mutant allele (f). Heritable variation is variation in traits / alleles / genes that can be passed on to the offspring. This means that the mutation for that variation must have occurred in the sex cell (gametes). Non-heritable variation comes from environmental effects on the somatic cell and cannot be passed on. This means that the mutation for that variation must have occurred in the somatic cell.	Accurately defines OR describes: • mutation • phenotype • genotype • genetic variation • heritable OR non-heritable.	• Explains the difference between phenotype and genotype, **using clear examples**. • Explains the difference between heritable and non-heritable variation. • Explains the difference between somatic and gametic mutations.	• Comprehensively discusses how genetic variation has occurred by linking key terms / ideas together (e.g. chromosomes, genes, alleles, DNA, phenotype, and genotype) within the context. • Comprehensively discusses genetic variation, which must reference the mutation of the F to f allele occurring in the gametic cell (for it to be passed on to future generations in the population) and

	The initial / original mutation for the CFTR gene must have occurred in the gametic cell because it has been passed on for many generations within the population.			**not** in the somatic cell.

N1	N2	A3	A4	M5	M6	E7	E8
ONE evidence point at Achievement.	TWO evidence points at Achievement.	THREE evidence points at Achievement.	FOUR evidence points at Achievement.	TWO evidence points at Merit.	THREE evidence points at Merit.	ONE evidence point at Excellence.	TWO evidence points at Excellence.

N0 = No response; no relevant evidence.

Question Two	Evidence	Achievement	Achievement with Merit	Achievement with Excellence
(a)(i)	FF (homozygous dominant) or Ff (heterozygous)	• Identifies **both** genotypes correctly.		
(ii)	Individual 4: FF (homozygous dominant) or Ff (heterozygous) Individual 5: ff (homozygous recessive) Individual 7: Ff (heterozygous)	• **Two of three** individuals correct.		
(b)	1 and 2 must be heterozygous (Ff) since 7 is unaffected, meaning that 7 must have **inherited** at least one dominant F allele from either parent. 1 and 2 also have affected children (5 and 6), therefore proving both are **heterozygotes** as they must have each passed on an f allele for a child to inherit cystic fibrosis. Both 7 and 8 must also be heterozygous, since they are unaffected and have two children who are both affected by cystic fibrosis. This means that each of them must have passed on a recessive f allele for a child to inherit cystic fibrosis. Punnett square crosses between 1 and 2 (or 7 and 8): 	• Identifies the genotypes of 1 and 2 (heterozygous). • Identifies the genotypes of 7 and 8 (heterozygous).	• Explains why 1 and 2 must be Ff, by referring to offspring (e.g. since 1 and 2 must each have at least a recessive f allele to pass on). • Explains why 7 and 8 must be Ff, by referring to their offspring (11 and 12) (e.g. since 7 and 8 must each have at least a recessive f allele to pass on).	• Comprehensively discusses the genotype of 7, using evidence from the parents (1 and 2), their offspring (5 and 6), and the offspring of 7 and 8, (11 and 12).

	F	**f**
F	FF	Ff
f	Ff	ff

(c)	4 and 10 are either FF or Ff. Both have unconfirmed / unknown genotypes. 4 is unaffected, therefore must have an F allele. 4 could have inherited an f allele from the heterozygous parents, hence could be Ff or FF. Since the offspring of 4 are unaffected, 9 and 10 could either be FF or Ff as well. If 3 and 4 have another child, and the child has cystic fibrosis, that must mean both 3 and 4 are heterozygous. Tracking the genetic relationship is important because 4 has a family history of cystic fibrosis. It would be beneficial to determine if 4 is FF or Ff. If 4 and 3 are Ff, it may mean that they have a chance to have a child who may inherit cystic fibrosis / carrier. This may impact on their decision to have more children. If 4 is Ff, it would be good to know if any of her offspring are carriers. This information may help her offspring (9 and 10) decide if they have children of their own in the future. This could involve developing targeted treatments / healthcare options for affected individuals. It would also help the advancement of medical knowledge of the disease; how it is passed on, etc. This can help improve future treatments.	• Identifies that 10 can be Ff or FF. • Identifies that 3 and 4 must be Ff if they have a child with cystic fibrosis. • Describes the purpose for genetic tracking.	• Explains why 4's genotype cannot be confirmed by referring to their offspring (9 and 10) • Explains a purpose for tracking cystic fibrosis in this family (3 and 4).	• Discusses the purpose of identifying the genetic relationship for the genotype of 4, including reference to 4's parents, their offspring, potential future offspring.

N1	N2	A3	A4	M5	M6	E7	E8
ONE evidence point at Achievement.	TWO evidence points at Achievement.	THREE evidence points at Achievement.	FOUR evidence points at Achievement.	TWO evidence points at Merit.	THREE evidence points at Merit.	ONE evidence point at Excellence.	TWO evidence points at Excellence.

N0 = No response; no relevant evidence.

Question Three	Evidence	Achievement	Achievement with Merit	Achievement with Excellence
(a)	Meiosis is a process of cell division producing four genetically non-identical daughter cells. Crossing over is when homologous chromosomes exchange genetic material, producing new unique combinations of alleles. Independent assortment is when homologous chromosomes line up in the middle of a cell in a random order before separating into gametes. These processes mean that there is a chance the f allele is passed on to the gametes and thus remains in the gene pool. Sexual reproduction is a biological process where offspring are produced through the combination of genetic material from two parent organisms. Fertilisation is the fusion of two gametes (sperm and egg). This means that even if both parents do not have cystic fibrosis, they may be carriers (of the recessive f allele) and have a chance of passing this on to their offspring. Non-random mating (selective mating) refers to mating / sexual reproduction that has not occurred due to chance, rather each mate has 'chosen' the other for a particular reason (e.g. physical or genetic traits). This could happen in small populations where there is a higher chance of two heterozygotes mating / inbreeding / mating with close relatives / second cousins.	• Accurately defines or describes meiosis. • Accurately defines or describes sexual reproduction. • Accurately defines or describes non-random mating. • Accurately defines or describes fertilisation. • Identifies that crossing over or independent assortment occurs during meiosis. • Identifies that two unaffected parents / carriers can produced affected offspring.	• Explains meiosis (includes a description of the effects of either crossing over or independent assortment). • Explains how sexual reproduction may cause carriers to pass on the recessive f allele to their offspring. • Explains how non-random mating (in small populations) can lead to a higher chance of the allele being passed on in the population.	• Comprehensively explains how meiosis, sexual reproduction, and non-random mating in a small population can lead to a higher chance of the f allele being passed on.

(b)	Population refers to a group of individuals of the same species living in the same area (capable of interbreeding). The f allele is disadvantageous in homozygous recessive individuals. It is also neither advantageous nor disadvantageous in a heterozygous individual. Heterozygotes may have an increased resistance to certain infectious diseases (e.g. cholera or typhoid). If this advantage improves reproductive success, this will increase the survival chances of heterozygotes, allowing them to reproduce and pass on the f allele, therefore increasing the f allele in the population. OR In smaller populations, more heterozygotes may end up mating together more often, resulting in a higher frequency of the f allele. This is due to random sampling effects in a small gene pool. Due to the dominant allele (F) completely overriding / masking the recessive allele (f), it is possible for the number of heterozygotes in a population to increase (therefore increasing the number of recessive alleles) without the number of cystic fibrosis phenotypes to increase (e.g. if there are fewer FF genotypes but more Ff genotypes). With the increase in availability of gene tracking and genetic testing, it is possible that the recessive f allele will decrease in frequency over the coming generations. This could be more likely in wealthy countries with more access to this technology. The decrease would be because prospective parents, particularly those with cystic fibrosis in the family, may have embryos / offspring tested while they are still a small group of cells. This could lead to not implanting an embryo (in the case of IVF) or terminating a pregnancy if the recessive f allele was present in the genotype.	• Accurately defines population. • States that the f allele will increase due to heterozygotes / carriers. • Identifies the f allele as being disadvantageous. • Identifies the f allele as being neither advantageous nor disadvantageous in heterozygotes.	• Explains a logical scenario for the f allele to increase, decrease, or stay the same. • Explains a reason for the f allele to increase in the population, while having no increase in cystic fibrosis phenotype.	• Comprehensively discusses AND clearly justifies why an increase in the f allele in a population does not necessarily lead to an increase in phenotype frequency.

N1	N2	A3	A4	M5	M6	E7	E8
ONE evidence point at Achievement.	TWO evidence points at Achievement.	THREE evidence points at Achievement.	FOUR evidence points at Achievement.	TWO evidence points at Merit.	THREE evidence points at Merit.	ONE evidence point at Excellence.	TWO evidence points at Excellence.

N0 = No response; no relevant evidence.