SUPERVISOR'S USE ONLY

1

92022

920220

Draw a cross through the box (☒) if you have NOT written in this booklet

+

NZQA | Mana Tohu Mātauranga o Aotearoa
New Zealand Qualifications Authority

Level 1 Chemistry and Biology 2025

92022 Demonstrate understanding of genetic variation in relation to an identified characteristic

Credits: Five

Achievement	Achievement with Merit	Achievement with Excellence
Demonstrate understanding of genetic variation in relation to an identified characteristic.	Explain genetic variation in relation to an identified characteristic.	Evaluate genetic variation in relation to an identified characteristic.

Check that the National Student Number (NSN) on your admission slip is the same as the number at the top of this page.

You should attempt ALL the questions in this booklet.

If you need more room for any answer, use the extra space provided at the back of this booklet.

Check that this booklet has pages 2–12 in the correct order and that none of these pages is blank.

Do not write in the margins (////). This area will be cut off when the booklet is marked.

YOU MUST HAND THIS BOOKLET TO THE SUPERVISOR AT THE END OF THE EXAMINATION.

Excellence

TOTAL 22

Page 1

QUESTION ONE: Chromosomes, genes, alleles, and DNA

The cystic fibrosis transmembrane conductance regulator (CFTR) gene in humans is responsible for mucus production in the human body.

The normal allele (F) is dominant over the mutant allele (f) in humans. Individuals who are homozygous recessive will develop cystic fibrosis, a condition where too much mucus is produced, which can cause difficulty breathing.

Figure 1: Location of the CFTR gene on a pair of chromosomes

(a) Explain the relationship between chromosomes, genes, alleles, and DNA. Include examples in your answer.

DNA is a double helix structure containing the genetic code for an organism. Chromosomes make up the rungs of the double helix structure of DNA, and are located inside the nucleus of the cell. They are made up of proteins and a single molecule of DNA, containing the genetic code for an organism. Genes are sections of DNA and chromosomes which code for a particular trait. Alleles are alternative versions of genes, which create different versions of the trait. For example, the gene coding for CFTR in humans contains alleles which determine how much muscus an individual will produce (e.g. excessive amount or normal amount).

(b) (i) Define the term **mutation**.

A mutation is a permanent change in the DNA base sequence which can result in the formation of a new protein and therefore a new allele and therefore a new trait.

(ii) Discuss how genetic variation has occurred in the CFTR gene.

In your answer, include:

- the difference between phenotype and genotype
- the difference between heritable and non-heritable variation.

In context of the CFTR gene, a mutation has occured resulting in a permanent change in the DNA base sequence of an individual. This has created a new mutant allele, which codes for the specific trait of producing too much mucus and having cystic fibrosis. A genotype is a combination of alleles resulting in the gene for a specific trait. A phenotype is the physical expression of the genotype. The mutant allele (f) coding for cystic fibrosis is recessive, which means that it will only show up in the phenotype of an invidiual when both recessive alleles are present in the genotype. However, when a dominant allele (F), which in this case codes for not having cystic fibrosis, is present in the genotype, it will mask the effects of the recessive allele regardless of if one is present or not. Therefore, in order to have the cystic fibrosis trait, an individual must be homozygous recessive (meaning both alleles in their genotype are recessive) with the genotype of ff.

Genetic variation has occured in the CFTR gene due to a mutation which introduced a new allele, in this case the mutant allele coding for cystic fibrosis. Because the mutant allele is recessive, it can be carried by individuals without appearing in their phenotype, so they won't produce excess mucus. The mutant allele however, can be passed down to offspring.

Genetic variation is differences between the DNA base sequences of individuals within a population. Due to the introduction of a new allele, differences between the genes and DNA base sequences of individuals has occured, resulting in different genotypes and phenotypes of individuals, therefore introducing genetic variation and new traits. Heritable variation is when these new alleles and traits can be passed down to the offspring of individuals through sexual reproduction and meiosis, where the gametes of two individuals fuse and their offspring gets a mix of their genetic material. Mutations can lead to heritable variation if their mutant alleles can be passed down to offspring. This can only happen when the mutation occurs in the gametes during meiosis. In context of the CFTR gene, inhertiable variation has been introduced because individuals are able to pass the mutant allele coding for cystic fibrosis down to their offspring, even if it does not always apppear in the phenotype of an individual, meaning the individual will not neccessarily get cystic fibrosis. Non-inhertiable variation is when mutations do not occur in the gametes, and cannot be passed down to offspring. For example, if a mutation were to occur in the somatic cells of an individual due to errors due mitosis (DNA replication), that may result in a change to the individual's phenotype, but they will not pass it down. These types of mutations can occur due to environmental factors. For example, if an individual worked in an area where they always inhale smoke, they may develope a lung cancer overtime which can cause them difficulty breathing similar to the way cystic fibrosis does. However, they would not be able to pass their lung cancer to their offspring because they developed that due to mutation from environmental factors, and the mutation did not occur in the gametes.

In context of the CFTR gene, heritable variation has occured due to the mutation producing the mutant allele coding for cystic fibrosis. Because the mutation created a new allele, this results in differences in the genes and DNA base sequences of an individual with the mutant allele compared to someone without, overall created variation. And because the mutation occured in the gametes, the mutant allele can be passed down (though not always shown in the phenotype of the individual with the mutant allele), leading to more individuals with the differences in their DNA base sequences as a result, and overall leading to heritable genetic varitation.

Source (adapted): https://stock.adobe.com/335315205, https://stock.adobe.com/763873950

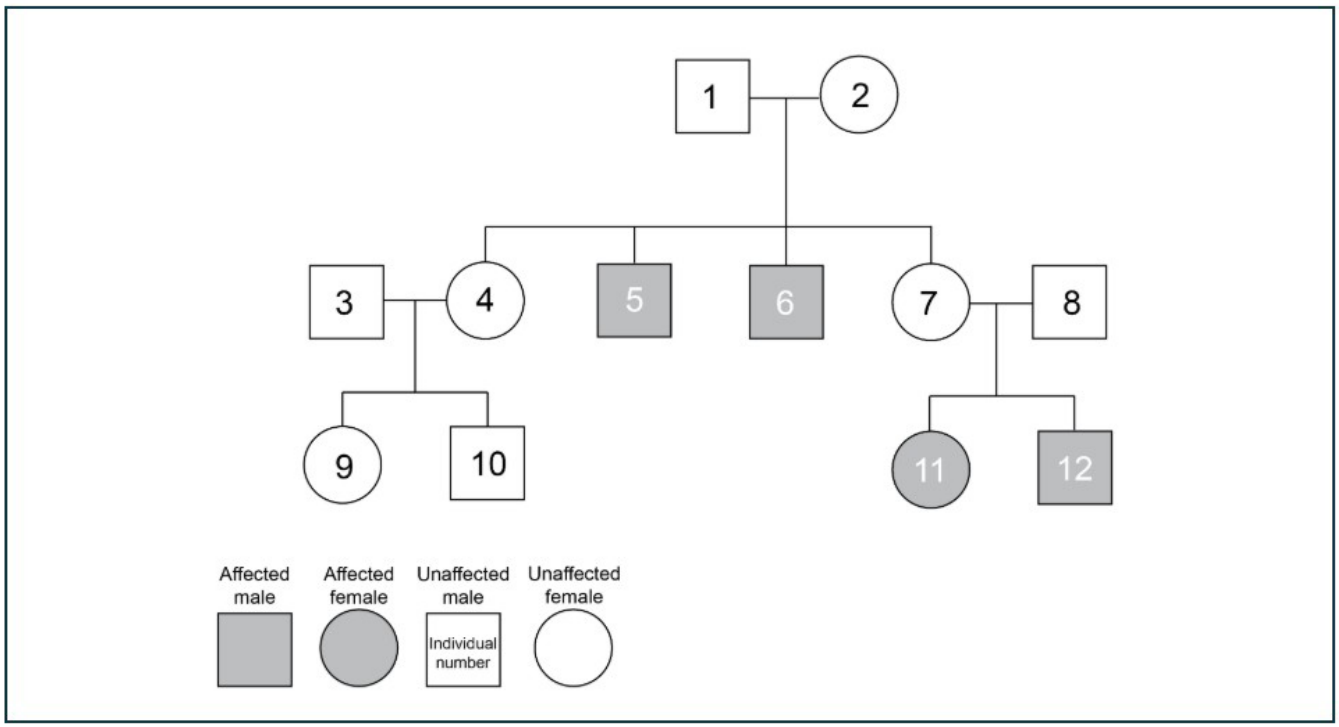
Page 2

QUESTION TWO: Tracking the cystic fibrosis allele

Cystic fibrosis is a heritable genetic disease in humans. It is caused by a recessive allele (f). The normal allele (F) is dominant over the mutant allele (f).

Figure 2 is a pedigree chart tracking the inheritance of cystic fibrosis in a family.

Figure 2: Inheritance of cystic fibrosis



(a) (i) What are the possible genotypes of an individual who **does not** have cystic fibrosis?

FF, Ff

(ii) Fill in the table below with the genotype(s) of the numbered individuals from Figure 2.

	Genotype		
Individual 4	FF	*or*	Ff
Individual 5	ff		
Individual 7	Ff		

(b) How are scientists able to confirm the genotype of individual 7 from Figure 2?
Support your answer using evidence from both the parents of individual 7 **and** the children of individuals 7 and 8.

Type into the boxes below to use as Punnett squares to support your answer.

	F	f
F	FF	Ff
f	Ff	ff

	F	F
f	Ff	Ff
f	Ff	Ff

Parents 1 and 2: Ff, Ff
Individual 7: Ff
Individual 8: Ff
Children 11 and 12: ff, ff

Homozygous is when both alleles in a genotype are the same. Homozygous recessive means they are both recessive alleles ff. Homozygous dominant means they are both dominant alleles FF. Heterozygous means the genotype contains both a dominant and recessive allele.

The mutant allele coding for cystic fibrosis is recessive (f), meaning it will only appear in the phenotype of an individual if both recessive alleles are present in the genotype. Therefore, to have cystic fibrosis an individual must have the genotype of ff. They must be homozygous recessive. For this to occur, there must be at least one recessive allele in the genotypes of both parents (as shown in the first punnet square). If one individual does not have at least one recessive allele in their phenotype, none of the offspring will be homozygous recessive (as shown in the second punnet square).

The allele coding for not having cystic fibrosis is dominant (F), meaning it will always appear in the phenotype of an individual when present in the genotype, it will mask the effects of the recessive allele if one is present. Therefore, to not have cystic fibrosis an individual can have the genotypes (combination of alleles) of either Ff or FF. They must be either heterozygous or homozygous dominant. For this to occur, there must be at least one parent with one dominant allele in their genotype.

During sexual reproduction, meiosis and the crossing over of alleles occurs. The genetic material of 2 parents combine, leaving their offspring with a combination of their alleles. In context of individual 7, both parents (1 and 2) are unaffected and do not have cystic fibrosis. This means they could have the genotypes of either FF, or Ff. However, their offspring 1 and 2 offspring are affected. Because the only possible genotype to be affected is with ff, and both parents are unaffected, the parents must have the genotypes of Ff and Ff, which is the only genotype that allows them to remain unaffected, while having the risk that their genetic material may combine to produce the genotype of ff, allowing them to have children with cystic fibrosis while they both do not. At the same time, two of their offspring are unaffected (individuals 7 and 4). Which means they could have a phenotype of either FF or Ff due to the combination of their parent's alleles from which they both have the Ff genotype.

Both parents 7 and 8 are unaffected. However, both of their offspring are affected. The only possible genotype to be affected is ff, but since 7 and 8 are unaffected they must have either FF or Ff. Since both their offspring are affected they must both have the ff genotype. To create this genotype while being unaffected parents, the parent's genotypes must be heterozygous, containing both a recessive and dominant allele. Both parents 7 and 8 must have the genotype of Ff, because in order to have affected offspring they must be homozygous recessive, and in order to be homozygous recessive both parents must have at least one recessive allele in their genotype (as shown in punnett 1). Therefore individual 7 can be proved to have the genotype of Ff.

(c) Discuss the purpose of identifying the genetic relationship for the genotype(s) of individual 4 from Figure 2.

In your answer, consider:

- the possible genotype(s) of individual 4 by referring to individual 10
- the purpose for tracking the cystic fibrosis allele in this family in relation to individual 4.

Individual 4 could have either the genotype of FF, or Ff, as they are an unaffected individual and the unaffected trait comes from the dominant allele F.
Individual 10 (offspring of 4 and 3) is also unaffected, and could have either the genotype of FF or Ff.

The purpose of identifying the genetic relationship for the possible genotypes of individual 4 is to know if individual 10 has the possibility of passing on the recessive allele if they have their own offspring. Individual 4's parents both have the genotypes of Ff, and since individual 4 is unaffected they cannot have the genotype of ff, therefore that leaves only Ff, Ff, and FF as a possibility. There is around a 66.67% chance that individual 4's genotype contains the recessive allele f coding for cystic fibrosis. Tracking the cystic fibrosis allele in this family would allow individual 4 to know if they might have passed on their recessive allele her two offspring, including individual 10. By doing this, individual 10 can be prepared that they may have an offspring with cystic fibrosis in the rare case that their partner also has this mutant allele, and their offspring gets ff as a combination of alleles from both their parents. Individual 10 can prepare for this by knowing if their parent individual 4 has this recessive allele or not.

Page 3

QUESTION THREE: Continuation in the population

The normal cystic fibrosis transmembrane conductance regulator (CFTR) allele (F) is dominant over the mutant allele (f). Homozygous recessive individuals will develop cystic fibrosis. Homozygous dominant and heterozygous individuals will not be affected.

(a) Explain how meiosis, sexual reproduction, and non-random mating can contribute to the continuation of the recessive allele (f) in the population.

Meiosis is the crossing over of and exchanging of alleles that occurs between two individuals during sexual reproduction. Meiosis is also the random assortment as alleles match up randomly and swap/cross over. Sexual reproduction is the fusing of two individual's gametes fuse which produce an offspring with a unique combination of both their parent's genetic material. These can contribute to the continuation of the recessive allele (f) in the population by allowing the mutant allele to be passed down to offspring. During the crossing over and exchanging of alleles, the recessive allele can be matched up and assorted randomly so that it ends up in the genotype of the new offspring. There, it can continue to be passed on throughout the population as more individuals breed and end up with new combinations of alleles, possibly containing the recessive allele (f) coding for cystic fibrosis.

(b) Discuss how the CFTR mutant allele (f) remains in the population.

In your answer, consider:

- if the occurrence of the recessive allele (f) will increase, decrease, or stay the same over time
- why an increase in the frequency of recessive alleles (f) in a population does not always lead to an increase in the frequency of the cystic fibrosis phenotype.

Overtime, the CFTR mutant allele (f) will increase in a population. As there is only a 25% chance that two unaffected individuals containing the recessive allele will pass it on and create an offspring who is homozygous recessive and has cystic fibrosis, but there is a 75% chance that the individual will not be affected, but still have the allele, there are going to be more individuals within a population with this recessive allele overtime, but not necessarily more individuals with cystic fibrosis. Individuals who are affected will have trouble breathing, and may die eariler and not reproduce, or they may decide not to reproduce for the sake of not risking to pass their condition to their offspring. However, more individual's within a population will have this recessive allele unknowingly, as they do not have cystic fibrosis. This is because they only need to have one dominant allele present in their genotype to mask the effects of the recessive mutant allele in their phenotype, meaning they will not have cystic fibrosis so long as they are not homozgyous recessive (with 2 recessive alleles). As the mutant allele does not show up in their phenotype, the individual does not have cystic fibrosis and will likely reproduce, with a chance of their mutant allele being passed on to their offspring again. Their offspring will now carry the mutant allele, but will most likely not have cystic fibrosis as their other parent would have to contain at least one of the mutant alleles for there to be just a 25% chance of that individual becoming homozygous recessive and having cystic fibrosis. So overtime, as individuals within a population reproduce, the mutant allele will likely be passed on to offspring, and passed down once again to their offspring, over generations until the recessive allele has increased its occurance within a population. However, this increase in the frequency of recessive alleles in a population will not necessarily lead to an increase in the frequency of the cystic fibrosis phenotype, as there is a much more slim chance of an offspring being homozygous recessive which is the only genotype possible to have cystic fibrosis. In order for that to happen both parents of an offspring would have to have at least one recessive allele in their phenotype, and the chance of their offspring being homozygous recessive with cystic fibrosis is still only 25%. So if only assuming only unaffected parents reproduce, there is either a 0% or 25% chance of their offspring having cystic fibrosis, which means the vast majority of a population in which the recessive allele has an increased occurance will mostly be unaffected and not have cystic fibrosis. Assuming that the little population who ends up being homozygous recessive and having the cystic fibrosis phenotype will be less likely to reproduce compared to those affected, due to not wanting to pass it on, or possibily earlier death, or just not having the capabilities to look after little ones due to their condition.

Excellence

Subject: Chemistry and Biology

Standard: 92022

Total score: 22

Q	Grade score	Marker commentary
One	E8	The candidate discussed clearly the difference between phenotype and genotype, and heritable and non-heritable variation using clear examples from the stimulus information. The candidate also demonstrated clear understanding of the relationship between chromosomes, genes, alleles, and DNA, and used clear examples to support their response.
Two	E7	This is a comprehensive response that discussed how genotypes of individuals from a pedigree chart can be confirmed, using key biological terms, and linking it to the stimulus information. The candidate provided a comprehensive response for the purpose of tracking an identified characteristic (cystic fibrosis) in individual 4, with respect to generational outcomes.
Three	E7	This is a thorough response that explained how meiosis, sexual reproduction, and non-random mating can contribute to the continuation of the recessive allele in the population. It also provided a comprehensive response on how the mutant allele remains in the population. The candidate made clear links to relevant biological ideas, confidently used biological terminology, and provided insight. Logical examples were used to support the discussion.