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

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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.

Merit

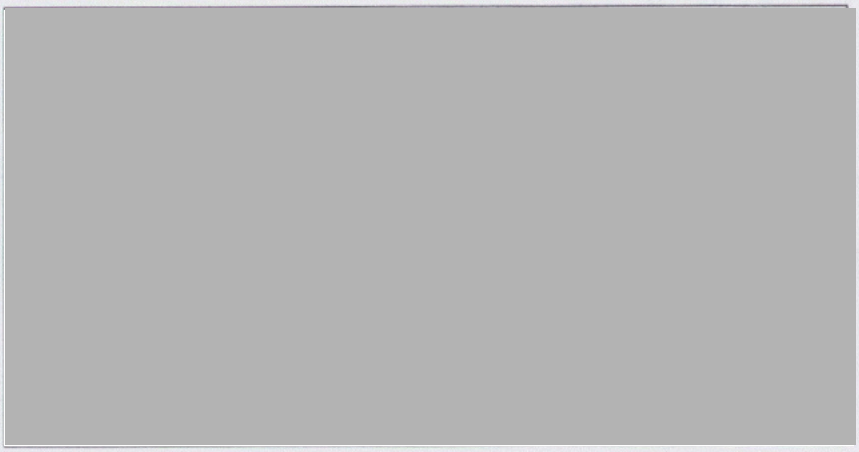
TOTAL 17

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 carries genetic information which is packed into chromosomes. Chromosomes are found in the nucleus of cells. Genes are sections of DNA that code for specific traits. An allele is one of two or more versions of a gene. You inherit two alleles for each trait, they then decide ~~your trait~~ what trait you convey. In this case the gene is the cystic fibrosis transmembrane conductance regulator. It is found on a chromosome. There are two types of alleles. The mutant allele (f) and the normal allele (F). ~~Depending on what you inherit~~ The alleles you have depend on what you inherit.

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

A mutation is a permanent change to the DNA sequence. This can be caused by an error in DNA replication or by mutagens.

(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. ✓

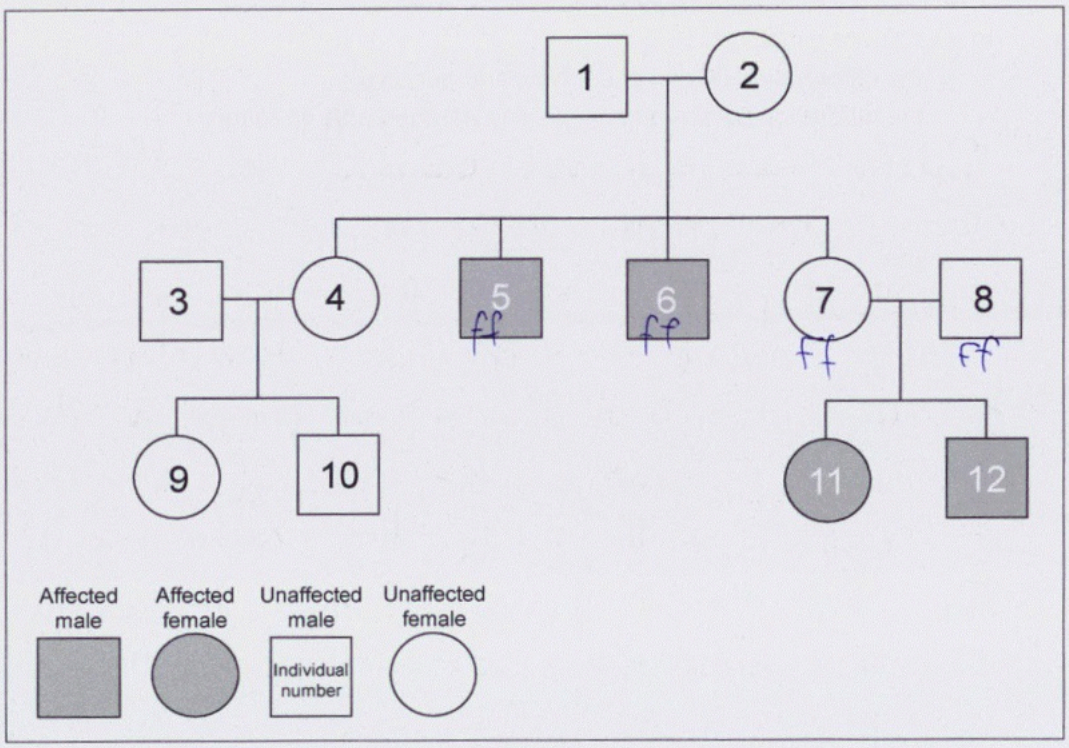
Genetic variation has occured in the CFTR gene. The mutant allele has led to more variety in genotypes. Genotype refers to the genetic makeup of an organism. Phenotype refers to the physical traits of an organism. This is decided by genotype but can be impacted by the environment. The mutation that occured is heritable. This means it can be passed down through generations by sexual reproduction. Non-heritable variation cannot be passed down as it occurs in somatic (body) cells. The CFTR mutation occured in a germetic cell as it can be passed down. The mutation caused a new allele to form. This led to a new genotypes and phenotypes. The heritable variation caused by this is not good as it leads to an increased risk of developing cystic fibrosis. The change to the DNA sequence caused the mutant allele to form which led to genetic variation in the CTFR gene.

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 (homozygous dominant) or Ff (heterozygous)

(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 (homozygous recessive)		
Individual 7	Ff (heterozygous)		

(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.

Use Punnett squares to support your answer.

1

	F	f
f	FF	Ff
f	Ff	ff

2

7

8

	F	f
F	FF	Ff
f	Ff	ff

Scientists can confirm the genotype of individual 7 through both her children and siblings. Individual 7 has two siblings affected by cystic fibrosis which means both parents (Individuals 1+2) must be carriers of the disease. The first punnett square shows that there is a 50% chance Individual 7 has the ~~genotype of~~ heterozygous genotype. Individuals 7's children are both affected by cystic fibrosis which means both parents must be carriers for both children to have the genotype of homozygous recessive. We inherit one allele from each parent which means both parents have to carry the allele.

(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.

The purpose of identifying the genotype of ~~gene~~ individual 4 is to track the cystic fibrosis allele. Individual 4 is unaffected which means she carries the dominant (normal) allele. Both of her children are also unaffected but she could still be a carrier as she has affected siblings. Her genotype could be Ff (heterozygous) or FF (homozygous dominant). By identifying her genotype he could

Answer space continues on the next page ➤

understand the risk of her children being carriers of the mutant allele. By tracking the gene, scientists can understand the genetic relationship. This could lead to solutions on how to prevent passing on the mutant allele and lead to early discoveries. Here tracking is used to track diseases and alleles through generations. If her son (Individual 10) ended up being a carrier this could lead to more offspring inheriting cystic fibrosis. Tracking the gene could help those at risk of spreading the allele more aware and could lead to early discoveries and treatment plans.

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.

Sexual reproduction is a biological process in which a new organism is created through the fusion of gametes. Meiosis is a cell type of cell division that halves the number of chromosomes. This creates gametes which in males is sperm and in females eggs (ovum). These processes result in organisms inherited half of their chromosomes from each parent. This creates genetic variation but also means that ya are more likely to inherit the cystic fibrosis allele (f). as Genotypes are decided by the alleles you inherit. By inheriting one from each parent you are more likely to get the recessive allele. Non-random mating can also contribute to the continuation of the recessive allele (f) because even knowing the genotype of two individuals doesn't ensure the offspring won't inherit the mutant allele (f) unless both parents are homozygous then dominant.

Question Three continues on the next page ➤

(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.

The CFTR mutant allele (f) remains in the population due to sexual reproduction and it being inheritable. The occurence of the recessive allele will likely increase. This is due to many people being carriers of the allele. The heterozygous genotype is very common which leads to offspring either inheriting just one mutant allele or both which results in cystic fibrosis. Although the number of recessive alleles (f) might increase that doesn't mean an increase in the frequency of the cystic fibrosis phenotype. To get cystic fibrosis you have to inherit the mutant allele from both parents which results in a homozygous recessive phenotype. Even if both parents are carriers this is uncommon due to the mutant allele being recessive. The mutant allele will continue to remain in the population as we continue to reproduce leading offspring to inherit the mutant allele. It is highly likely the allele will increase in frequency due to its ability to be passed through generations. It will increase as its inheritable and the heterozygous genotype is very common. Although the cystic fibrosis phenotype

won't necessarily increase the frequency of the mutant allele(f). will.

Acknowledgements
Material from the following source has been adapted for use in this assessment:

Figure 1 https://stock.adobe.com/335315205, https://stock.adobe.com/763873950

Merit

Subject: Chemistry and Biology

Standard: 92022

Total score: 17

Q	Grade score	Marker commentary
One	M6	The candidate explained the difference between phenotype and genotype, and heritable and non-heritable variation using examples from the stimulus information. The candidate also explained the relationship between chromosomes, genes, alleles, and DNA, and used clear examples to support their response.
Two	M6	This is an in-depth response that explained 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 an in-depth response for the purpose of tracking an identified characteristic (cystic fibrosis) in individual 4, using clear examples linked to inheritance / their offspring, to support their response.
Three	M5	This is an in-depth response that explained how meiosis and sexual reproduction can contribute to the continuation of the recessive allele in the population. It also explained how the mutant allele remains in the population. The candidate used relevant biological ideas and provided clear examples to support their response.