SUPERVISOR'S USE ONLY

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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.

Achievement TOTAL 10

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.

The relationship between chromosomes, genes, alleles and DNA are very important. DNA is the overall thing that holds all our genetic information, this includes genes, alleles and chromosomes. Chromosomes are made of DNA and proteins and ~~code for a specific trait. Genes~~ carry the genetic instructions. Genes code for a specific trait and the allele is another version of a gene. They all work together.

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

Mutation is a permanent change in your DNA sequence.

(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.

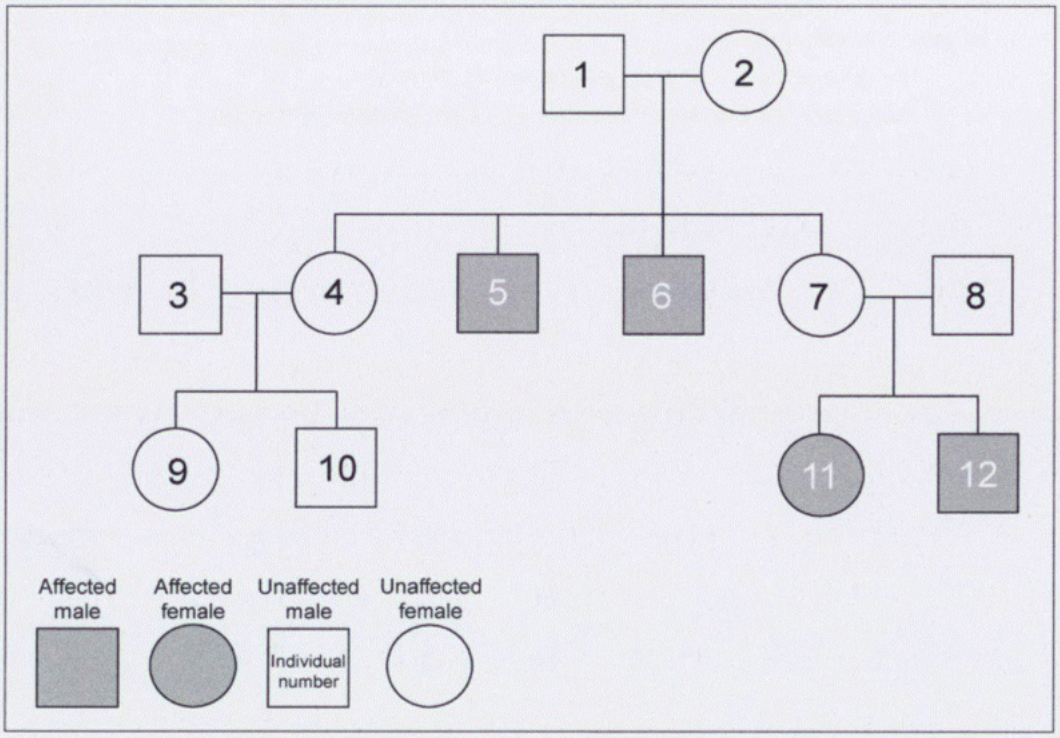
Genetic variation has occured in CFTR gene. The difference between the genotype and phenotype are important. A genotype is a combination of alleles that codes for a trait. A phenotype is the physical expression of this genotype, like blue eyes. That is the difference between them both. In this case you don't have the genotype of CFTR, then you don't have that physical trait. There is also a difference between heritable and non heritable variation. Hevitable is where the specific gene will be passed down many generations. We call this gametic this is because the gene is created in the sex cells. Non heritable is where you cannot pass a trait down. This is called Somatic this occurs in your own individual cells and cannot be passed on. CFTR is a heritable 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?

The possible genotypes would be, FF and 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.

Use Punnett squares to support your answer.

	F	f
F	FF	Ff
f	Ff	ff

	F	F
F	FF	FF
f	Ff	Ff

Scientists are able to confirm the genotype of Individual 7 by looking at her parents and her children. We use their information. Both of her parents are unaffected but still have off springs with the mutation. This doesn't confirm 7s genotype but we have a clearer understanding. We then look at her partner and her off springs. ~~Her and he~~ 7 and 8 are both unaffected ~~but~~ produce two off springs with the [continues on the extra space]

(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.

It is important to identify the genetic relationship for the genotype of individual 4. How we can do this is by looking at the results of 4's children. Individual 4 who is unaffected reproduces with Individual 3 who is also unaffected. 4 has siblings that have the mutation. ~~When 3 and 4 rep~~ Both of 4s parents don't have the mutation.

Answer space continues on the next page ➤

When 3 and 4 reproduced they had two off spring neither affected. We can't assume that individual 4 doesn't carry the heterozygous recessive gene. We just know that 4 ~~carry~~ carries a dominant allele. It is important for this family to track the CFTR gene since it isn't dominant. Any of the unaffected individuals could be carrying it, ~~but~~ but you wouldn't know because its recessive. They need to track it since it's a gametic mutation and generations down the line could still be affected.

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.

The process of meiosis is where a cell divides and splits up into 4 different cells, these cells are used for sexual reproduction. This process can contribute to the continuation of the recessive allele, since this trait is gametic it is passed down through the sex cells. If they chose not to randomly mate, and choose someone with CFTR. That would make the chances so much higher for the next generations to get it.

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 will remain in this population. The occurence now of this recessive allele is mediumly high, but I think as time goes on I think it will decrease. I don't think there will be huge drop of the people who have it, but I think it will be lower. It will still be in the population just not as common. ~~Fdb~~ If there was an increase in the frequency in recessive alleles that doesnot always mean the frequency of the cystic fibrosis phenotype will increase. ~~Just because~~ The allele for CFTR is recessive this means people can still carry the allele as well as the dominant unmutant allele. That means it is not expressed because the mutant is recessive.

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

Extra space if required.
Write the question number(s) if applicable.

QUESTION NUMBER

Question 2 b

mutation. This means that individual 7 has to carry the mutant allele. Individual 7 doesn't express this because her genotype is heterozygous dominant, Ff. We know this by using the information of 7s parents and children.

Achievement

Subject: Chemistry and Biology

Standard: 92022

Total score: 10

Q	Grade score	Marker commentary
One	A4	The candidate described the difference between phenotype and genotype, and heritable and non-heritable variation using examples from the stimulus information. The candidate described / defined the necessary key terms (chromosomes, genes, alleles, and DNA).
Two	A3	The response described how genotypes of individuals from a pedigree chart can be confirmed, using key individuals from stimulus information. The candidate described a purpose of tracking an identified characteristic (cystic fibrosis) in individual 4, using a simple example to support their response.
Three	A3	The response described how meiosis and sexual reproduction can contribute to the continuation of the recessive allele in the population. It also described why an increase in the frequency of the mutant allele does not always lead to an increase in the frequency of the cystic fibrosis phenotype.