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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 2 Biology 2024

91157 Demonstrate understanding of genetic variation and change

Credits: Four

Achievement	Achievement with Merit	Achievement with Excellence
Demonstrate understanding of genetic variation and change.	Demonstrate in-depth understanding of genetic variation and change.	Demonstrate comprehensive understanding of genetic variation and change.

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 2–16 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.

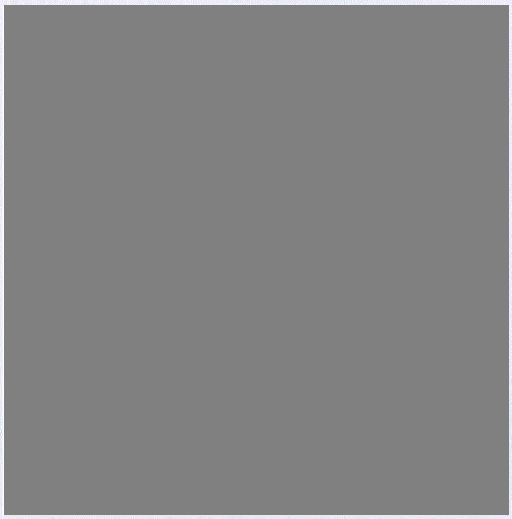
Merit

TOTAL 16

QUESTION ONE: Cats

Some domestic cats exhibit a complete dominance pattern in coat colour. The allele for black fur (B) is dominant over the allele for brown fur (b). The gene for tail length is not linked and is located on a different chromosome from the coat colour gene. The allele for long tails (L) is dominant to the allele for short tails (l).

A cat that is homozygous for both black fur and long tail is crossed with a cat that is homozygous for both brown fur and short tail.



Domestic cats.

(a) Specify the genotype of individuals in the F1 generation produced by this cross.

BbLl

(b) Use the Punnett square below to show:

- the F1 gametes resulting from the cross **and**
- the possible genotypes within the F2 generation of cats.

F1 gametes

F1 gametes	BL	Bl	bL	bl
BL	BBLL	BBLl	BbLL	BbLl
Bl	BBLl	BBll	BbLl	Bbll
bL	BbLL	BbLl	bbLL	bbLl
bl	BbLl	Bbll	bbLl	bbll

(c) Give the expected phenotype ratio resulting from this cross, indicating the phenotype each value represents.

9 Black long tail
3 black short tail
3 brown long tail
1 brown short tail

Some cats have fur colour that is an example of co-dominance AND sex linkage. In cats with orange fur, phaeomelanin (orange pigment) completely replaces eumelanin (black or brown pigment). This gene is located on the X chromosome. The orange fur allele is (F) and is co-dominant with non-orange (f). Males can typically only be orange or non-orange (black, brown, etc.).

Sex determination in cats is the same as humans.

Female cats can have orange fur, fur without any orange (black, brown, etc.), or have tortoiseshell fur (see image on the right), in which some parts of the fur are orange and others are non-orange.

Some cat diseases are known to be sex-linked as well. Male cats have been found to be more susceptible to recessive, sex-linked diseases than female cats.

Female black tortoiseshell.

(d) Evaluate the inheritance patterns of cats to include complete dominance, co-dominance, and sex-linkage.

In your answer, refer to the examples above and include a discussion of:

- the two patterns of dominance
- why only female cats can have tortoiseshell fur colour
- the similarities and differences of recessive and dominant sex-linked genes.

Codominance is when both alleles are equally dominant to one another, and both will be expressed equally if heterozygous, like the tortoiseshell fur. Complete dominance is when the dominant allele will completely mask over the recess, so if dominant allele is present, dominant phenotype will always be expressed. Codominance will have 3 different phenotypes, ~~dominance phenotype~~, mixed of both equally, or

recessive. When both alleles are present, Females have tortoiseshell fur because females have codominant.
XX chromosomes, and the F allele can occur on both chromosomes. However, male cat have XY chromosomes, and F can only be on one of the chromosome, X. Hence male cats can only be orange or non-orange, not both if recessive f allele is inherited by male cats on X chromosome, orange fur wont show, But for female XX, since the orange allele is completely dominant over non-orange (B or b) alleles. However, if both XX have F and f allele for female, Codominance will express the mixed colours. For recessive sex linked alleles, it will be masked by the other allele, and won't be expressed. This is why if male cats inherit the recessive allele that only X can carry, the phenotype won't be expressed. Contrastly, dominant alleles will always show, so if male cat inherit domint F allele, orange can will be expressed. Similarly, if dominant allele for disease is inherited, male cat will suffer from the disease as Y chromosome alleles cant mask it. But female cats can avoid diseases since they have 2 X chromosomes, and allele can be masked. Since F and f are codominant and only located on X chromosomes, heterozygous Ff will show both colours. But

male cat can only inherit one allele on their X chromosome, so codominance can't occur.

QUESTION TWO: Takahē

The flightless takahē (*Porphyrio hochstetteri*) has special cultural, spiritual, and traditional significance to Ngāi Tahu, the iwi from New Zealand's South Island. Ngāi Tahu value takahē as a taonga (treasure), and they continue to act as kaitiaki (guardians) of the takahē, working alongside the Department of Conservation / Te Papa Atawhai (DOC).

Takahē in a protected colony.

Genetic analyses and fossil records show that takahē were restricted to isolated areas in the north-western South Island at the height of the last ice age, approximately 29 000–19 000 years ago. As the climate warmed, takahē shifted their distribution, migrating to eastern and southern regions. The takahē in the north-west South Island became locally extinct. Pressures from hunting, introduced predators, habitat destruction, and competition for food led to their decline and an extreme genetic bottleneck.

After being presumed extinct for nearly 50 years, the takahē was famously rediscovered in 1948. The rediscovery of the takahē led to New Zealand's longest-running, endangered species programme. For more than 70 years, measures to protect and increase numbers of takahē have included predator control, captive breeding, and island translocations (moving small populations of birds to offshore islands).

Ongoing genetic analyses have found that introduced island populations of takahē have significantly lower levels of genetic variation than the main Fiordland population. The island population also has significantly different gene frequencies, with some alleles becoming fixed (with no variability in the gene pools) on the island sanctuaries.

Discuss the decline in genetic diversity in the takahē, with reference to the information provided.

In your answer, include discussion of:

- the terms **population bottleneck**, **founder effect**, and **genetic drift**
- how the genetic diversity of the gene pools of the takahē have been impacted by these processes
- why the reduced genetic diversity from island translocation is a problem for the takahē population and how this may be improved in the future.

~~population bottle~~ Genetic drift is the random change in allele frequency of a gene pool. It can include population ~~bottleneck~~, which is the large reduction of individuals ~~of~~ in a population such as habitat destruction or predation for the Takahē. Founders effect is when a small number of individuals establishes into a new population that is not representative of the original population. ~~This ear~~ For example, a ~~sma~~ number of takahē migrated to eastern and southern (E and S) regions from ~~AA~~ north-western ~~sout~~ areas. The small number of individuals establishing a new population will affect the original ~~gen~~ allele frequency of the gene pool as alleles are leaving the gene pool. The new population gene pool in E and S regions are not representative of the original as they might not carry all the original alleles. This decreases the genetic variation for ~~both~~ populations, as allele variety will decreases in the gene pool. Bottleneck effect occured in the extreme decline of Takahē due to factors such as hunting, food competetion and many more. The death of the large numbers of Takahē reduces genetic variation as alleles may be lost or fixed, and allele frequency will be changed. The alleles carried by Takahē will leave the gene pool if an individual is killed, and this decreased genetic variation, especially if a large number of individuals are killed, like the Takahē. Since genetic drift is up to chance, the allele frequency is also

changed randomly, which may result in the loss of a beneficial allele. This will decrease survival rate of offspring, and result in lower genetic variation and small populations. A small population and low genetic variation is affected by genetic drift more than larger population. It increases the risk of fixed / lost alleles as there are less individuals carrying a variety of alleles. The death of takahē, or when they migrate / translocate will have a larger proportional impact on the allele frequency. This is why low genetic diversity has a large impact on translocation, as the Takahē's that are moved will have even lower genetic variation, impacting greatly the allele frequency of the original (already small) population, and the new translocated population. The low genetic variation and small population may also increase risk of inbreeding which will affect the offspring. A larger population has higher chance of random mating. So, with the low genetic variation, inbreeding can occur in the translocated population. As alleles are fixed due to the small population and genetic variation, alleles can only be introduced if mutation occur or new alleles migrate in a gene pool. This can be prevented in the future by allowing the population to grow larger first, reducing risk of fixed / lost alleles, and inbreeding. It will also increase genetic variation. After, translocation can occur, and these factors that affect allele frequency will be reduced.

QUESTION THREE: Lethal alleles

In 1907, Erwin Baur carried out research on the snapdragon plant, *Antirrhinum majus,* and studied the condition known as 'aurea', in which some plants produced golden leaves instead of green leaves. In this plant, the golden-leaf allele (G) is dominant to the green-leaf allele (g). When crossed with its own type (aurea × aurea), Baur observed a 2:1 phenotype ratio of golden:green-leafed plants, instead of the expected 3:1 ratio in the offspring.

By carrying out a number of test crosses, Baur concluded that all of the surviving golden-leafed plants were heterozygous. Homozygous dominant (GG) aurea plants lacked normal chlorophyll development and never survived.

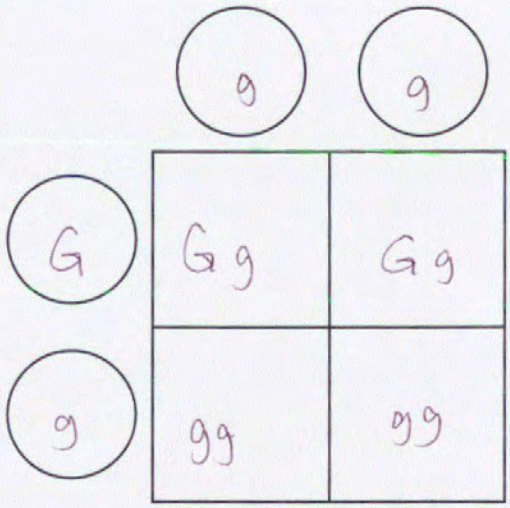
Baur is now recognised as the first scientist to discover lethal alleles in a plant, although they had already been recognised in animals, including humans.

Antirrhinum majus, snapdragon in bloom.

(a) Describe what is meant by the term **lethal allele**.

Lethal alleles are alleles that can result in death of an organism if both alleles are present, like the gold leaf allele.

(b) In the space below, show the Punnett square for a test cross between a green-leaf plant and a surviving, golden-leaf plant:



Genotype ratio: 1:1

Phenotype ratio: 1:1

(c) This lethal allele gives a dominant, non-lethal phenotype in the heterozygote. However, we say that the lethality (ability to prevent survival) is recessive in the snapdragon, even though the colour phenotype is dominant.

Using the information provided, discuss why this snapdragon allele must be recessive for lethality and why dominant lethal alleles are rare but can be found in some adult populations, including humans.

In your answer, include discussion of:

- the terms and meaning of dominant and recessive alleles
- why the snapdragon's allele must be recessive for lethality and how the test cross shows this
- why dominant lethal alleles are rarer than recessive ones, but can sometimes exist in adult organisms.

Dominant alleles will always mask over recessive if it is present in a population. ~~tet~~ Snapdragons lethality must be recessive, as the snapdragon is alive. Lethal alleles result death in the snapdragon due to the lack of chlorophyll required for survival. This means that although the golden phenotype of G is dominant, and will be expressed if it is present, over the green phenotype, this G allele must be recessive in lethality to ensure survival.

If the lethality of the golden allele is dominant, there will be no flowers with the golden phenotype, as the lethality will mask over and kill the flower. Since there are flowers with golden phenotype, we know that the golden allele is present, but lethality is being mask over, resulting in the survival. The test cross above also proves that lethality of G allele is recessive, as the phenotypic and genotype ratio are the same, meaning that the golden allele is expressed and survived. For G lethality is dominant, there will only be green leaf phenotypes as all golden leaves are killed due to lack of chlorophyll. However, if both lethal alleles are present, the recessive genotype will be expressed, and will kill the flower. The can be shown by Baur's cross between two gaden leaf flowers. Both are carries of the lethal allele, so offspring can inherit both, have GG. This offspring will not survive, which is why the actual phenotype ratio and expected ratio is different, as no GG will survive.

Dominant lethal alleles rare such as the gold leaf is rare for humans as the expression of the lethal phenotype will decrease survival of humans. The lethal phenotype can only be present if it doesn't affect survival, such as the gold colour of leaves. If the lethal phenotype is expressed, it is usually deadly, and the person won't be able to survive until it reproduces, so the allele won't be passed on.

This is why dominant lethal are rare, as most offspring won't inherit it. However, it is still there, as some individuals may be able to survive and pass on this lethal allele. Lethal alleles that are recessive for phenotype expression are more common, as a person could be carrying a lethal allele and not be affected by it. The person will then be able to survive and pass down the lethal alleles, and offspring can inherit it, which is why it remains in a gene pool. Dominant lethal allele will affect the person even if they are carries, hence it is rare. A person withe lethal allele also MUST be heterozygous, as is they are dominant with both lethal alleles, they won't survive, hence, lethality is recessive, and will be masked by normal alle.

Merit

Subject: Biology

Standard: 91157

Total score: 16

Q	Grade score	Marker commentary
One	M5	The response explains why female cats can have tortoiseshell fur, while male cats can only have orange or non-orange fur. The explanation effectively incorporates the concepts of both complete and co-dominance. Additionally, the response correctly identifies and uses the sex chromosome genotypes for both male and female cats.
Two	M5	The response correctly states the definitions of both the bottleneck effect and genetic drift, explaining their impact on genetic diversity.
Three	M6	The response successfully incorporates the concepts of 2:1 versus 3:1 ratios and a test cross to explain how lethal alleles are inherited. The candidate has also used the test cross to clarify why the snapdragon lethal allele is recessive. Additionally, the candidate explains that some dominant lethal alleles can exist into adulthood and describes how they can be inherited and established within a population.