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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.

Excellence

TOTAL 19

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.

B - black fur / L - long tails
b - brown fur / l - short tails.

BL → bl

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.

BbLl

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

BL B1 bL

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

9; Black fur - long tails, 3; Black fur, short tails, 3; brown fur - long tails, 1; brown fur - short tails.

F - orange = complete dominance

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.). ↑ only co-dom.

50/50 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.

F - f = co-dominant.

(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.

this is between the orange fur allele (F) and non orange (f)

The first pattern of dominance, within cats is co-dominance, co-dominance is where there are two 'dominant' alleles, and when there is a heterozygous individual, both phenotypes will be displayed (each cell can display a different dominant gene). The second pattern is complete dominance, where the phaeomelinin (orange pigment) completely masks the eumelinin, meaning any cat with the orange pigment allele will display the orange phenotype

this is move common in males. Sex linkage, is where there are alleles coding for a particular trait carried on the sex chromosomes, however this doesnt change the 50/50 posibilities for each sex. It just means that only females (XX) chromosomes ~~are~~ can have tourtise-shell fur. Because the gene for black or brown pigment is only found on the X chromosome, which males only have one of (XY). This brown or black pigment is a ressesive allele, meaning it is only produced in the phenotype when its present twice. As its only found on the X chromosome, only female cats can display it as they must have 2 ressesive genotype (black or brown) on 2 X chromosomes → eg; XX → both ressesive trait. Males can not however because even if they have one ressesive allele, X, the other is a Y chromosome, meaning it will mask the ressesive trait and then will not be displayed in the phenotype. ∴ only females that have the genotype Ff will have tourtouseshell colours - f = ressesive and F = co-dominant - ressesive wont be masked, but it will still have the orange pigment aswell as the black/brown ∴ turtouseshell. However a males doesnt have 2 X chromosomes so ressesive (eumelinin) will always be masked.

~~A dominant sex linked gene is~~ A ressesive sex-linked gene likely decreases genetic diversity because it is not often ~~liable~~ expressed. Must have two

coppies (XX) chromosomes to be visable in phenotype. Sex linked deiseises can be very haumful to a population. 'Male cats have been found to be move suceptible to sex-linked deiseses than females' meaning this deisiese is likely found on the Y chromosome, and ∴ only inhevited by males. on, move likely is a co-clominant allele which is only sometimes expressed in males, and vavely in females. Dominant sex-linked genes ave likely to always be inhevited, unless on the Y chromosome - then only males would ALWAYS get that allele. However a vessesive sex-linked gene only gets expressed in the phenotype if it is displayed twice (XX) chromosomes, so vessesive genes ave move likely to be on the X chromosomes, (would hever be expressed on Y chromosome as X would always mask it). The similavities is that they ~~ave~~ both dont effect the gender vatio of offspving, and could both have potential positive and negitive conceqences too offspring.

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.

The bottleneck effect / population bottleneck is the rapid decline in a population due to a catastrophic event or human intervention. We see this in the takahē population through the original population ↓ numbers being so dramatically decreased due to hunting, introduced predators, habitat distruction and competition for food. The loss of individuals ~~many~~ mean that allele frequencies / gene pool variation will change. The remaining population is unlikely to have a representitive gene pool - with all the same alleles, as the population before the catastrophic events ∴ decreasing genetic diversity. The founders effect is where a small proportion of a population migrate / become isolated, ~~which~~ ~~mean~~ and they establish there own population, with a different gene pool. Again this new-smaller population's gene pool is unlikely to be representitive of the original population and there is likely less genetic variation due to alleles getting lost. We see this in the takahē when predator control, captive breeding and translocation as well as migration after the climate warmed, where the takahē have moved to new locations (migration) or been forced to move (translocation) and have established new gene pools / populations that would be less genetically diverse, as alleles have likely been reduced / lost. Genetic drift is the change of allele frequencies over time, due to random chance. It effects the now much smaller takahe populations

as random casualties could eliminate alleles completely from a gene pool and ∴ severley decrease genetic variation. Whereas a larger population such as the original takahē population wouldnt be so heavily affected by small casualties ∴ genetic drift wont be heavily affected either. Bottleneck effect, founder affect and genetic drift would have severley limited the genetic diversity of the takahēs gene pools. However, as explained above, the introduced island populations of takahe will have lower genetic diversity as its a smaller population than the main fiordland population. ∴ ~~genetic drift is~~ lost alleles due to any 3 of those processes mean that a smaller populations gene pool is more affected. Island translocation has lead to very low genetic diversity, with some alleles becoming fixed (only one version of a gene for a particular trait). This may lead to interbreeding within the takahē population on the islands/ may be a result of. This means even fewer recombinants of alleles would be possible ∴ decreasing genetic diversity even further, ∴ is problamatic. It also means if selection pressures such as the previous increased temperature or habitat distruction/predation/habitat change, the individuals may not have fit alleles to survive these changes. It could be improved in the future through further relocation, into optimal conditions, or they could re-locate the island sanctuary birds back into the mainland population, to breed with and ∴ re-establish a more genetically diverse gene pool for the future.

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.

↑ lethal (not expressed)

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.

↑ lethal allele

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.

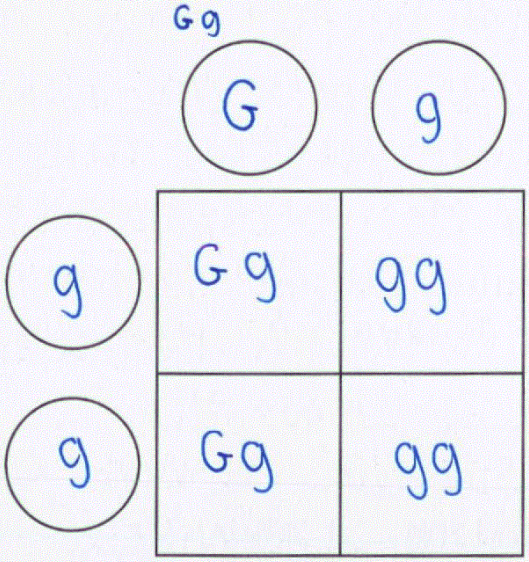
G golden
g green

Antirrhinum majus, snapdragon in bloom.

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

A lethal allele is an allele that when expressed in the genotype, is never observed in the phenotype, it generally codes for a ~~pro~~ dis-functioning protein that harms / eventually kills the organism. A lethal allele is an alternate version of a gene that will kill the organism it inhabits. Unless it is ressesive and with a dominant allele ∴ isn't always expressed / doesn't always kill.

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



← surviving golden plant.

Genotype ratio: 2; Gg, 2; gg

Phenotype ratio: 2; gold leaf; 2 green leaf.

(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 are alleles that will always be expressed in the phenotype, eg; GG. A recessive allele is an allele that is only displayed in the phenotype if it is homozygous, or else a dominant allele will mask it. eg; Gg. The snapdragons lethal allele must be recessive because it is not expressed in the above punnit square/ all organisms crossed have survived. However if the allele was dominant, there would be lethal

a much lower rate of survival, and the phenotypic ratio would not be the same as the genotypic, as individuals would die. However because it is recessive, there must be two lethal alleles present for the offspring to die; GG, where the chlorophyl development means they wont survive. Therefore the alleles 'lethality' is recessive, as it cannot fully ~~control~~ the organisms survival. It must be present recessive for lethality, otherwise it would be dominant and ∴ would always kill the offspring ∴ no lethality / no ability to prevent survival, like the recessive ~~de~~lethality allele can. For a dominant lethal allele to become established in a gene pool (homosapiens) ~~of~~ it needs to become inherited. Dominant lethal alleles are rare because individuals that have them aren't likely to make it to the age where they can reproduce / pass on the gene. Because they are likely to die before reproductive age - although with some exceptions such as huntingtons disease, sometimes a adult dominant lethal allele will exist in a gene pool/organism. However, recessive alleles are more frequent, because they can be carried without being expressed. For example ~~two~~ can individuals may have the recessive allele 'a', and ~~can~~ would still live a happy normal life if the recessive lethal allele is masked by a

dominant non-lethal allele. These individuals are far more likely to survive and reproduce, ∴ passing on this allele to further generations ∴ making them far more common, and maybe even established within a gene pool. It is only when ~~an~~ ~~organ~~ two heterozygous individuals who carry the recessive allele reproduce to make a homozygous recessive offspring that the recessive allele would kill the offspring.

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

QUESTION NUMBER

91157

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

Page 2
https://stock.adobe.com/207291020

Page 3
https://en.wikipedia.org/wiki/Cat_coat_genetics

Page 6
https://www.doc.govt.nz/nature/native-animals/birds/birds-a-z/takahe/

Page 10
https://stock.adobe.com/324607740

Excellence

Subject: Biology

Standard: 91157

Total score: 19

Q	Grade score	Marker commentary
One	M5	The response accurately identifies the sex chromosome genotype for a female cat, and explains how the presence of two different alleles, F and f, lead to tortoise-shell fur colour. The Punnett square has been completed correctly and describes the phenotype ratio.
Two	E7	The response effectively uses the context of the takahē provided in the question to discuss the effects of genetic drift and the founder effect on genetic variation. It successfully creates links between the two processes, discussing their implications on the takahē population as the different mechanisms of change impact the population over many years.
Three	E7	This response uses the concepts of dominant and recessive alleles to explain how a dominant lethal allele can persist in a population, contrasting it with the more common occurrence of recessive lethal alleles. The inclusion of Huntington's disease as an example demonstrates a comprehensive understanding of these concepts.