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91157



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## Level 2 Biology, 2015

### 91157 Demonstrate understanding of genetic variation and change

9.30 a.m. Monday 16 November 2015

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 space for any answer, use the page(s) provided at the back of this booklet and clearly number the question.

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

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

Excellence

TOTAL

21

ASSESSOR'S USE ONLY

## QUESTION ONE: BLACK ROBINS



<http://nzbirdsonline.org.nz/species/black-robin>

Introduced species such as cats and rats caused the Chatham Island black robin (*Petroica traversi*) population to plummet to five individuals in 1980. Due to intensive conservation efforts, the species now has over 250 individuals in the gene pool.

- (a) Describe the term gene pool.

// A gene pool is all of the alleles present within a population.  
A population being 1 species of organism living in a defined  
area //

- (b) Explain how genetic drift affects the black robin's gene pool.

// As the black robin's population is very small genetic drift  
will have a significant effect on the gene pool. Genetic drift is  
the loss ~~or gain~~ of alleles in a gene pool due to chance and not  
selection. The range of alleles (genetic biodiversity) of the black  
robin population will not be large thus if one individual is  
eliminated from the population if its alleles are not eliminated  
completely from the gene pool they will certainly decrease in  
frequency dramatically. //

- (c) Female black robins usually lay eggs inside their nests. However, conservationists found some birds laid eggs on the rims of nests, where the eggs could not survive. So, they pushed the eggs back into the nests where they could be incubated and hatch successfully. However, this selection pressure from humans caused the rim laying allele to increase to 50% in the black robin population. They decided to stop pushing eggs back into the nests to prevent the behaviour from spreading throughout the population. In 2011 only 9% of the population laid eggs on the rims of nests.



Nest showing egg laid on rim.  
[www.math.canterbury.ac.nz/~r.sainudiin/preprints/plos\\_br\\_preprint.pdf](http://www.math.canterbury.ac.nz/~r.sainudiin/preprints/plos_br_preprint.pdf)

Discuss why some female black robins lay eggs on the rims of nests, while most lay eggs inside the nests, and how humans affected this behaviour.

In your answer include:

- a description of what allele and allele frequency mean
- an explanation of what selection pressures are, and how they affect natural selection
- a discussion of natural selection using the black robin egg laying example
- a discussion of why the rim laying behaviour increased with human intervention, then decreased once the intervention stopped.

*//An allele is an alternative form of a gene, e.g. the gene for egg laying has two forms: laying on the rim or laying in the centre of the nest. The allele frequency is the number of times an allele is present in a population's gene pool (or how frequent it is). Selection pressures are ~~environmental~~<sup>human</sup> pressures which alter the environment's natural selection process. In the case of the black robin natural selection selected that rim laying black robin allele would not survive. If humans had not interfered then the eggs that had been laid on the rim of nest and so may have inherited this allele from their parent would not have survived to pass on this allele, and so the allele would not have survived and thus the black robin population would have grown stronger - only ~~people~~<sup>birds</sup> with the centre laying allele would have survived, thus those individuals with this allele, would have survived to reproduce. The fittest individuals would have survived and so passed on their*

**There is more space for your answer to this question on the following page.**

Alleles. As populations generally have more individuals than the environment can sustain competition ~~can~~<sup>will</sup> ensue for survival. Because humans interfered with the natural selection process by placing rim laying eggs back into the centre of the allele where they could be incubated properly, the organisms of these eggs survived. They will have inherited the allele from their parents of rim laying thus when they survived to grow up they laid their eggs on the rim of the nest - increasing the frequency of the rim laying allele <sup>1 to 50%</sup> in the population. When humans stopped interfering with the natural selection process the birds that were laid in eggs on the rim of their nests died. This means they did not survive to reproduce, and so over time the rim <sup>laying</sup> allele decreased in frequency in the gene pool. The individuals with the fittest-centre laying-allele survived, passing this on to their offspring, so a stronger black robin population resulted.

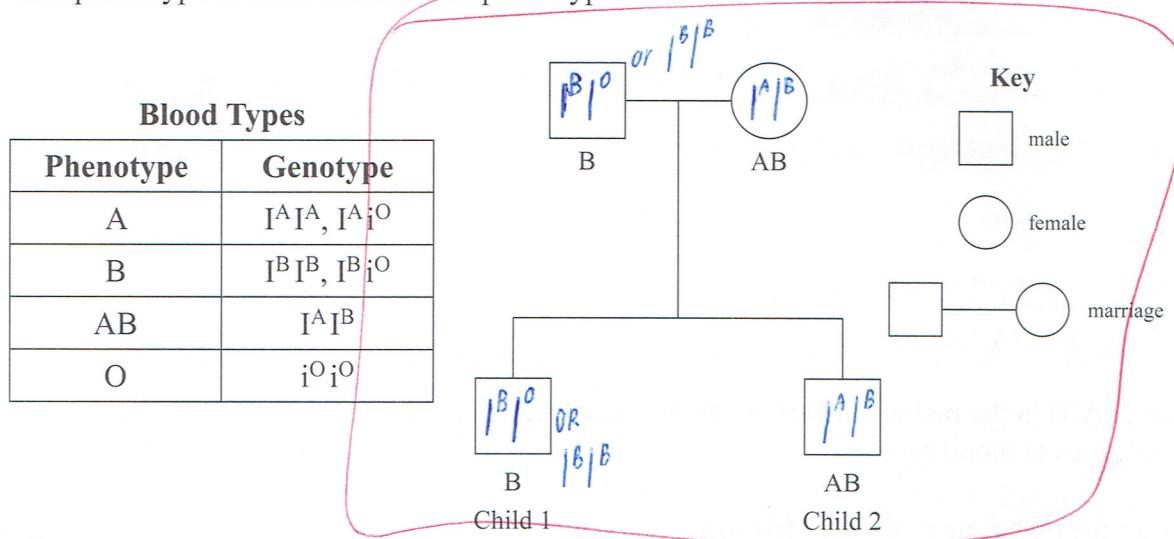
## QUESTION TWO: BLOOD TYPE

There are multiple alleles that determine a human's blood type. These are known as  $I^A$ ,  $I^B$  and  $i^O$ . Alleles  $I^A$  and  $I^B$  are dominant over  $i^O$ . However, when  $I^A$  and  $I^B$  are inherited together, they show co-dominance.

- (a) Describe what multiple alleles are.

*// Multiple alleles arise when there is more than 2 alleles for a gene, although only 2 of these alleles will be inherited by an individual in the genotype to be expressed in the phenotype. //*

- (b) The pedigree chart below shows the two children and their phenotypes that result from a male with phenotype B and a female with phenotype AB.



Explain why Child 1 has two possible genotypes while Child 2 has only one possible genotype.

You may use diagrams in your answer.

*// Child 2 has 1 possible genotype as the only genotype that codes for AB blood type is  $I^A I^B$ .  $I^A$  and  $I^B$  are codominant and so are expressed in equal amounts creating the genotype  $I^A I^B$ . Child 2 can not have Child 1 can have 2 possible genotypes however as 2 genotypes result in the phenotype of blood type B.  $I^B I^B$  is the homozygous dominant form of the genotype, and as both alleles are B the phenotype of B is inherited in blood type. However, if*

*There is more space for your answer to this question on the following page.*

the child has the genotype  $I^B I^0$ , they will still express the phenotype of B blood type. This is because the allele  $I^B$  is completely dominant of the ~~gen~~ allele  $I^0$ , so in the ~~ge~~ heterozygous genotype where  $I^B$  and  $I^0$  are both present  $I^B$  will always be expressed as only 1 copy of that allele needs to be present in the genotype to be expressed. In Child 2's case however  $I^B$  and  $I^A$  are co-dominant so in this genotype  $I^B$  is not completely dominant over  $I^A$  as it is over  $I^0$ . ~~because~~  $I^A$  and  $I^B$  are equally dominant and so both alleles are expressed in the phenotype creating the blood type  $I^A I^B - AB$ .

- (c) Child 2 (AB) in the pedigree chart on the previous page has children with a female having homozygous O blood type.

Discuss the inheritance of their offspring.

In your answer include:

- the possible phenotypes AND genotypes of the offspring
- an explanation of the difference between dominance and co-dominance
- a discussion of why none of their children will have the blood type O or AB.

You may use diagrams in your answer.

If ~~child~~ individual AB-genotype  $I^A I^B -$  has a child with an individual with the genotype  $I^0 I^0$  then the possible offsprings' <sup>genotypes</sup> that may result are as followed:  $I^A I^0$  and  $I^B I^0$ . Resulting phenotypes will be expressions of the  $I^A I^0$  and  $I^B I^0$  genotypes in the forms of blood types A and B. Dominance occurs when an allele is always completely dominant over another. Only 1 form of that allele needs to be present in a heterozygous genotype for it to

be expressed - i.e.  $I^A I^0$ ,  $I^A$  is always dominant over allele  $I^0$ , so when it is present in a heterozygous genotype the blood type phenotype will always result. Co-dominance arises when both alleles in the genotype are equally dominant. No one allele is dominant over the other, thus both are expressed equally in the phenotype - i.e.  $I^A I^B$ ,  $I^A$  and  $I^B$  are equally dominant and so are expressed in equal amounts in the phenotype-creating AB blood types.

The individuals  $I^A I^B$  and  $I^0 I^0$  can never produce a child with the genotype  $I^0 I^0$ , and thus phenotype blood group O, as  $I^0$  allele ~~does not have~~ has to be inherited from each parent. Individual  $I^A I^B$  does not have an  $I^0$  allele to donate to the offspring, and as allele  $I^0$  is recessive to  $I^A$  and  $I^B$  it needs to be present twice in the genotype (homozygous recessive) in order to be expressed in the phenotype  $I^0 I^0$ . The individual  $I^A I^B$  can only donate an  $I^A$  or  $I^B$  allele, and as both of these are dominant to  $I^0$  in the genotype  $I^A I^0$  or  $I^B I^0$  blood group A or B will always be shown.

Blood type AB can never result in an offspring as again for this to occur an  $I^A$  ~~or~~  $I^B$  allele from one parent and an  $I^B$  allele from the other would have to be inherited in order for the genotype  $I^A I^B$  to arise, and as  $I^A$  and  $I^B$  are equally dominant (co-dominant) the blood type AB results. Individual  $I^0 I^0$  can only ever donate an  $I^0$  allele, and this combined with child 2's either  $I^A$  or  $I^B$  will create the genotypes  $I^A I^0$  or  $I^B I^0$ .  $I^A$  and  $I^B$  are completely dominant to  $I^0$  thus will always be expressed and so the offspring can only ever have the phenotype of blood group A with the genotype  $I^A I^0$  or the phenotype of B blood group with genotype  $I^B I^0$ . Last Offspring will ~~not be~~ be heterozygous.

EF

### QUESTION THREE: COAT COLOUR

In 1905, Lucien Cuénot observed unusual ratios when studying inheritance of coat colour in mice. After mating two heterozygous yellow mice ( $Yy$ ), he observed that the offspring never showed a normal 3:1 phenotypic ratio. Instead, he always observed a 2:1 ratio, with two yellow mice for every grey mouse. He concluded that yellow coat colour ( $Y$ ) was dominant over grey coat colour ( $y$ ), and by using test crosses he showed that all his yellow mice were heterozygotes. However, from his many crosses, Cuénot never produced a single homozygous dominant yellow mouse.

Subsequently, it was confirmed that no homozygous dominant yellow mice were present because of a lethal allele.

[www.themouseconnection.org/t955-what-are-these-sooty-colors](http://www.themouseconnection.org/t955-what-are-these-sooty-colors)

$$Y = \text{yellow} \quad y = \text{grey}$$

- (a) Describe a lethal allele(s).

A lethal allele arises when a mutation causes a non-functional version of a protein to be produced. When a lethal combination of alleles are inherited the organism will die before or shortly after death due to the non-functional protein.

- (b) Discuss how Cuénot used test crosses to determine that all the live yellow mice were heterozygous.

In your answer include:

- a description of homozygous AND heterozygous
- an explanation of what a test cross is
- a discussion of how Cuénot used the test crosses to observe a 2:1 ratio (two yellow mice for every grey mouse), and determine that all live yellow mice were heterozygous.

You may use diagrams in your answer.

Homozygous refers to a genotype in which the two alleles present are the same - e.g.  $YY$ , homozygous dominant, and  $yy$ , homozygous recessive. Heterozygous refers to a genotype in which the two alleles present are different. In cases of complete dominance the dominant allele will always be expressed - e.g.  $Yy$  - heterozygous dominant,  $Y$  will be expressed. A test cross is a test to see whether or not an individual is pure breeding or not. The individual is breed/crossed with a homozygous recessive individual, in this way if any of individuals

If are produced which show the recessive characteristic then we know that the individual in question must be heterozygous for the individual to have inherited 2 of the recessive alleles (as one allele must be inherited from each parent).

	Y	y
y	Yy	yy
y	Yy	yy

Cuénat could confirm through test crossing that his mice were heterozygous as the recessive trait was shown in the offspring, showing that the recessive y allele must have been present in his mice. Due to lethal alleles all of the homozygous dominant rats died, thus this lethal combination of alleles when inherited caused a yellow rat to die. All surviving yellow mice must thus have a y allele present, and be heterozygous in order to pass this trait on to an offspring. ~~Ham & Cuénat cannot have any pure breeding yellow mice.~~

- (c) The genetic disease cystic fibrosis is caused by lethal alleles. An affected individual is homozygous recessive, however heterozygous individuals are carriers of the lethal allele. Lethal alleles are caused by mutations. The mutation for cystic fibrosis occurs in the gametes.

Discuss how mutations cause lethal alleles, AND why cystic fibrosis alleles remain in the population.

In your answer include:

- a description of what a mutation is
- an explanation of the difference between a gametic mutation and a somatic mutation
- a discussion of why the cystic fibrosis lethal allele remains in the human population.

A gene mutation occurs when a change to the base sequencing arises in DNA, resulting in a different amino acid being coded for, and thus a different protein. A protein formed then may have no function, a different function or an impaired biological function. Base sequencing can be mutated by 1 base being substituted for another or a base being inserted or deleted into the base sequencing resulting in a frameshift. A gametic mutation is a mutation that occurs in the ova in females or the sperm in males. A somatic mutation is one that occurs in body cells. The difference is that gametic mutations can be inherited / passed on as the ova or sperm with

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the mutation fuse to form the zygote which thus carries this mutated gene. A somatic mutation can not be inherited as the mutations occur in the body cells and <sup>their DNA is</sup> ~~these are not~~ passed on in reproduction. ✓

The lethal allele for cystic fibrosis still remains in the population today as the mutation must occur in the gamete cells. This means that the allele can be inherited by offspring. An individual can still survive if they only ~~&~~ have 1 allele for cystic fibrosis - they are heterozygous, thus the allele is still present. <sup>as individuals can survive</sup> If 2 parents have the allele for cystic fibrosis then an offspring could inherit the cystic fibrosis allele from both parents, resulting in them having the lethal combination of homozygous alleles, and thus not surviving! ✓

Excellence exemplar for 91157 - 2015			Total score	21
Q	Grade score	Annotation		
1	8	Correct definitions in part (a) (b) and (c). Provides a very good discussion integrating the egg laying behaviour of the Robin to Natural Selection as well as how the alleles for rim laying behaviour increased initially in the gene pool due to human intervention AND how the alleles decreased in the gene pool after human intervention had stopped.		
2	7	Provides an excellent discussion in Part (c) to discuss why the children cannot have blood type O or AB. Use of words like “equal amounts” for Co Dominance and link to Dominance as one allele being completely dominant over another ( without the recessive allele being mentioned). There is no mention of parental gametes and fertilisation in the answer. The grade was marked at E 7 than at E8.		
3	6	Provides correct definitions in (a) (b) and (c). Does not provide sufficient discussion in (b). Has used punnet square to show a test cross. However, there is no discussion as to why there is a deviation from the usual 3:1 to a 2:1 with experimental evidence. Has explained the difference between gametic and somatic mutations. The Cystic Fibrosis question was clearly discussed to a merit level as learner did not provide either an annotated punnet square to show 25% death or mention in the answer as to how only $\frac{1}{4}$ or 25% of the offspring from the homozygous genotype would be affected with Cystic fibrosis if 2 carriers of the lethal recessive allele produced a homozygous offspring.		