Mana Tohu Mātauranga o Aotearoa
New Zealand Qualifications Authority

Level 1 Chemistry and Biology RAS 2023

92022 Demonstrate understanding of genetic variation in relation to an identified characteristic

EXEMPLAR

Excellence

TOTAL 08

Chemistry and Biology 1.3 (92022)
Demonstrate understanding of genetic variation in relation to an identified characteristic
ANSWER DOCUMENT

Write your answers below:

Genetic variation is the difference between individuals within a population. It has different sources, which include sexual reproduction and mutation. Genetic variation is important for a species's survival, as differences among individuals means an increased chance that the species will survive through environmental change.

Genetic variation is the result of sexual reproduction and mutations. The physical appearance, phenotype, of an individual is determined by their genotype, their genetic information. A segment of DNA is called a gene, which codes for certain proteins. There can be variations of genes, alleles, which result in different phenotypes. An individual's alleles depend on their parents. Chromosomes, packages of DNA, are inherited. This means that an individual will inherit two versions of every allele, one paternal and one maternal. Fertilisation, where the egg and sperm cell fuse, results in randomly paired gametes with unique alleles. Before this, there is another source of variation. Meiosis involves a single cell dividing to create four unique gametes. They are unique because of three processes. Crossing over is a process where homologous chromosomes- of the same type- exchange information so that every chromosome will have a unique combination of alleles. On top of this, independent assortment aligns chromosomes randomly, and segregation ensures they are randomly separated into two nucleuses. These processes result in individuals having unique allele combinations.

On top of this, mutations can cause variation. These are changes in the base sequences within a genetic code, which alter protein production. They can be silent and not change anything, or they can be nonsense mutations, and cause change. Just one base pair change can cause a mutation. Mutations vary in their effect on the species. They can be harmful, neutral, or beneficial. As well as this, mutations can only be passed if they affect sex cells. Any changes to somatic - body - cells, will not be inherited by offspring.

Genetic variation is seen everywhere around the world. An example is Sickle Cell Anaemia. Within humans, there is a mutation, the sickle cell mutation, which leads to variation within the protein haemoglobin. Haemoglobin is a protein that carries oxygen around the body. The mutation causes misshapen proteins which impacts oxygen absorption. This disease reduces life expectancy. This allele has three phenotypes depending on the genotype. Homozygous dominant has no disease, homozygous recessive has the disease, and heterozygous genotypes have the sickle cell trait, which has no symptoms but allows that person to pass on the gene. This is why the mutated allele is continuing in the population, despite those with the disease not reaching child bearing age. Punnett squares are a good method of predicting the genotypes of offspring. While these are probability based, therefore not completely accurate,

they still provide an idea of the chances. For example, two carriers have a 50% chance of having carrier offspring, and 25% chance of both disease and no disease offspring.

Carrier/carrier			Carrier/No disease			Carrier/disease		
	R	**r**		**R**	**r**		**R**	**r**
R	RR	Rr	**R**	RR	Rr	**r**	Rr	rr
r	Rr	rr	**R**	RR	Rr	**r**	Rr	rr

The sickle cell carrier has been found to provide resistance to malaria. This advantage against malaria has led to the favourable trait (Sickle cell carrier) being found in high percentages in high malaria risk zones. This was found using gene tracking studies, such as DNA fingerprinting. This is because the people who carried the sickle cell allele were more likely to survive malaria, and continue to reproduce and pass on the mutation. This has led to the heterozygous genotype being common among people with an African background, but less so in other cultures. This shows the importance of genetic variation. By having diversity in the genotypes and phenotypes of people, the overall species responded well to malaria, a threat the environment posed.

The kakapo is another example. At one point, kakapo were incredibly endangered. Scientists used genetic information to decide how to save the species. It was important to have the kakapo's genetic information. This was because the information revealed that there was little variation between individuals. There was only one individual who showed genetic diversity. Scientists started a breeding program with the unique individual, because the lack of variation was hindering their survival. It was important to introduce variation to the population because diversity would better prepare them for environmental change, as the favourable trait would survive and reproduce, allowing for adaptation over time rather than extinction. Genetic data could inform scientists to breed individuals who would introduce more variety and help the species thrive. To maximise variety within kakapo and continue with their conservation, scientists should keep regular records of kakapo genetic information, so any variation or lack thereof can be noted. Genetic information should remain an important tool in the decision of which individuals to breed to promote more variation. By using punnett squares and pedigree charts, genotypes could be predicted and recorded, allowing for a better understanding of how future breeding programmes should be approached.

In conclusion, genetic variation is the differences within a population. It is a result of sexual reproduction and mutations. Variation allows individuals with favourable traits to survive and reproduce, aiding in long term survival. Therefore, genetic diversity is important for the survival of species. Two examples that show how genetic variation can be important are

sickle cell mutation, as the heterozygous allele provides malaria resistance, and kakapo, who almost died out from a lack of variation.

Excellence

Subject: Chemistry and Biology

Standard: 92022

Total score: 08

Q	Grade score	Marker commentary
(a)	E8	This response sits at Excellence level because the candidate has evaluated findings when genetic variation has been identified and tracked for the purpose of identifying genetic relationships. The candidate has also defined key biological terms, explained how mutation can be inherited, and has then applied this to the example of sickle cell anemia. They have used Punnet squares to show how genes can be tracked and particular alleles inherited. They have evaluated the usefulness of this information, and identified ways in which people may use it.
(b)		The candidate has evaluated the effectiveness of genetic tracking by linking the purpose of genetic tracking of kākāpō (the conservation effort) to the findings (the genetic differences between birds).