**Copy of BCC LogoBelmont City College**

**YR12 HUMAN BIOLOGY 2022 Unit 4**

**Task 6: Test – Variation, Mutation and Evolution**

**Time allowed: 55 minutes**

**Name: ANSWERS** Total Mark: /50

**Section One: Multiple Choice [10 marks]**

Place a ~~cross~~ through the selected letter.

1. A B C D 6. A B C D

2. A B C D 7. A B C D

3. A B C D 8. A B C D

4. A B C D 9. A B C D

5. A B C D 10. A B C D

**Section Two: Short Answer [25 marks]**

**Question 11 (10 marks)**

## Martha’s Vineyard is an island off the coast of the USA. During the 1700s and 1800s there were a large

## number of Deaf individuals within the population on the island.

## What effect was most likely to be responsible for the large numbers of Deaf individuals?

## (1 mark)

## **The Founder Effect (1)**

## Briefly explain how this effect would have caused the high number of deaf individuals on the island

## (4 marks)

|  |  |
| --- | --- |
| **Description** | **Marks** |
| * **Small (isolated) sample of original population** * **This sample is not genetically representative of original population** * **With some/more individuals carrying the genes for deafness** * **Restricted breeding/inbreeding among the population/restricted gene pool** * **Frequency of deaf gene maintained over time** | **Four points for 4 marks** |
|  | **Total 4** |

1. Suggest a factor that would have changed in the 1900’s to result in the reduced incidence of genetically-inherited deafness in the Martha’s Vineyard population.

(1 mark)

|  |  |
| --- | --- |
| **Description** | **Marks** |
| * **(increased) migration from the mainland to the island** * **interbreeding/gene flow with the mainland that increased breeding** | **Any one point for 1 mark**  **(max 1)** |
|  | **Total 1** |

1. Identify another example of a specific population with a high incidence of a genetically- inherited disease. State the population and the disease in your answer.

(2 marks)

|  |  |
| --- | --- |
| **Description** | **Marks** |
| **Any disease for 1 mark and matching population 1 mark**   * **Tay-Sachs disease in Ashkenazi/Jewish population** * **Achromatopsia in Pingelap population** * **Thalassaemia in Greek population** * **Sickle-cell anaemia in African population** | **Any two points for 1 mark each (max 2)** |
|  | **Total 2** |
|  |  |

1. A mutation is another type of evolutionary mechanism that can affect frequencies of alleles in populations. Explain the difference in consequences between a mutation occurring in a somatic cell and one occurring in a gamete (2 marks)

|  |  |
| --- | --- |
| **Description** | **Marks** |
| **Somatic affects only the individual (1), gamete is inheritable change (1)** | **2 marks** |

**Question 12 (5 marks)**

West Nile Virus (WNV) is currently on the rise in developed countries such as the USA, and is known to lethally infect humans. Studies have found that allele for the protein CCR5(+) and its mutated form CCR5∆32 are associated with WNV susceptibility. The table below shows the distribution of genotypes in patients with and without WNV in Northern America.

|  |  |  |  |
| --- | --- | --- | --- |
| Genotype: | +/+ | +/∆32 | ∆32/∆32 |
| Patients with WNV | 321 | 78 | 18 |
| Patients without WNV | 155 | 16 | 2 |

1. Using the data above, the allelic frequency of **CCR5∆32** homozygotes was calculated and is stated below:

Patients with **CCR5∆32** and with WNV: 0.04

Patients with **CCR5∆32** and without WNV: 0.01

Given that the percentage of **CCR5∆32 homozygotes** in North America is usually 1%, compare frequency of **CCR5∆32 homozygotes** in both populations studied. (2 marks)

**The frequency of CCR5∆32 homozygotes is much higher (1) in the group with WNV (1)**

1. Propose an explanation for the data, stating whether an advantage is present. (3 marks)

**The data suggests that being CCR5∆32 homozygous is a disadvantage (1), as it appears to make people with these alleles more susceptible to West Nile Virus (1). Therefore, carrying the CCR5+ allele is an advantage where West Nile Virus is present (1).**

**Question 13 (10 marks)**

Since World War I, mustard gas has been used as a weapon in chemical warfare. Within 24 hours of exposure to mustard gas, victims develop chemical burns, which appear as large blisters to the exposed skin and respiratory surfaces.

In 1940, Auerbach and Robson completed the first research into the other potential effects of mustard gas. They exposed *Drosophila* flies to mustard gas and then examined the chromosomal damage in the flies’ offspring over several generations. Their results showed a dramatic increase in the number of chromosomal mutations compared to the control group.

Najafi and others (2014) studied human victims 25 years after their exposure to mustard gas during the Iran/Iraq War. It found there were 122 different mutated genes in the respiratory pathways of the victims.

1. The studies described above outline how mustard gas has been shown to cause mutations.

Mustard gas can therefore be classified as a: (1 mark)

**Mutagen (1)**

1. Auerbach and Robson were studying chromosomal mutations. Describe **two** types of these

mutations that can occur in organisms. (4 marks)

|  |  |
| --- | --- |
| **Description** | **Marks** |
| Any 2 – 1 mark for naming, 1 mark for description | |
| Insertions – extra base pairs are added into the DNA/chromosome. | 1–4 |
| Duplications – a section of DNA/whole chromosome is repeated |
| Deletions – a section of DNA is lost/removed |
| Translocations – a section of one chromosome attaches to another chromosome |
| Inversions – a broken segment is reversed and reinserted into a  chromosome |
| Non-dysjunction – extra or missing chromosome |
| **Total** | **4** |

1. The 2014 study examined mutated genes. Describe how this type of mutation differs from a

chromosomal mutation. (2 marks)

|  |  |
| --- | --- |
| **Description** | **Marks** |
| Occurs a single gene loci/doesn’t involve the whole chromosome | 1 |
| Change in one (or a few) nucleotide bases/substitution, insertion, or deletion of nucleotide bases | 1 |
| **Total** | **2** |

1. Explain why Auerbach and Robson studied the offspring flies, not the parent flies, and what

information this gave about the type of mutations that occurred. (3 marks)

|  |  |
| --- | --- |
| **Description** | **Marks** |
| Any 3 of: | |
| Trying to see if mutation can be passed on/was passed generation to generation | 1–3 |
| Mutations had to occur in the germ cells/reproductive cells/mutation present in gametes of parents |
| Mutations were germline/not somatic mutations |
| Mutations should have been found in all cells of the offspring |
| **Total** | **3** |

**Section Three: Extended Response [15 marks]**

Sickle Cell Anaemia is a disease that occurs due to a gene mutation affecting the formation of haemoglobin, and therefore the shape of the red blood cells. This disease is one example of where *heterozygote advantage* can occur.

Define heterozygote advantage, then use the principles of natural selection to explain how heterozygote advantage occurs with Sickle Cell Anaemia.

***Up to 2 marks for:***

***Heterozygote Advantage : carrying one allele (1) for a deadly recessive trait is an advantage in some circumstances (1)***

Up to 13 marks for the following. Example must be linked with correct principle for marks to be allocated.

|  |  |
| --- | --- |
| ***Principle of Natural Selection*** | **Deadly Recessive Disease** |
| **Sickle Cell Anaemia** |
| *There is variation of characteristics within a species. (1)* | Some of the population has a recessive allele that causes sickle shape (sickling) of RBC. (1) |
| *More offspring are produced than survive and reproduce. (1)* | Offspring born with two sickling alleles (1) (homozygous recessive -ss) cannot adequately transport oxygen and die in childhood. (1) |
| *There is competition for survival and this is linked to environment. (1)* | Presence of malaria increases competition (selection pressure) (1) |
| *Individuals with characteristics most suited to the environment have more chance of surviving and reproducing. (1)* | Individuals carrying one sickling allele (heterozygous - Ss) do not have anaemia, and are also resistant to malaria (1). In areas with malaria, this is a survival and reproductive advantage.(1) |
| *Favourable characteristics are passed on to the next generation. (1)* | In areas with malaria the sickling allele is more likely to be passed on (1), as people without the allele (SS) are more likely to die from malaria. (1) |
| *The proportion of favourable alleles in the population gradually increases. (1)* | We see a greater allele frequency (1) for the sickling allele in areas where malaria is present. (1) |

**END OF TEST PAPER**