**Year 12**

**ATAR**

**Human Biology**

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| Name: |
| Teacher: |

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| **Task 8: Mutations and Gene Pools Test** | | | | Weighting 5% |
|  | Marks Received | Marks Available | Percentage | |
| Multiple Choice |  | 10 |  | |
| Short Answer |  | 25 |  | |
| Extended Answer |  | 20 |  | |
| Total |  | 55 |  | |

Time Allocated:

Reading time: 5 minutes

Working time: 55 minutes

**Year 12**

**ATAR**

**Human Biology**

**MULTIPLE CHOICE ANSWER SHEET**

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| Name: |
| Teacher: |

For each question, shade the box to indicate your answer. Use only a blue or black pen to shade the boxes. If you make a mistake, place a cross through that square then shade your new answer. Do not erase or use correction fluid/tape. Marks will not be deducted for incorrect answers. No marks will be given if more than one answer is completed for any question.

|  |  |  |  |  |
| --- | --- | --- | --- | --- |
| **1** | a | b | c | d |
| **2** | a | b | c | d |
| **3** | a | b | c | d |
| **4** | a | b | c | d |
| **5** | a | b | c | d |
| **6** | a | b | c | d |
| **7** | a | b | c | d |
| **8** | a | b | c | d |
| **9** | a | b | c | d |
| **10** | a | b | c | d |

**PART A: Multiple Choice Section [Total: 13 marks]**

1. A gene pool is

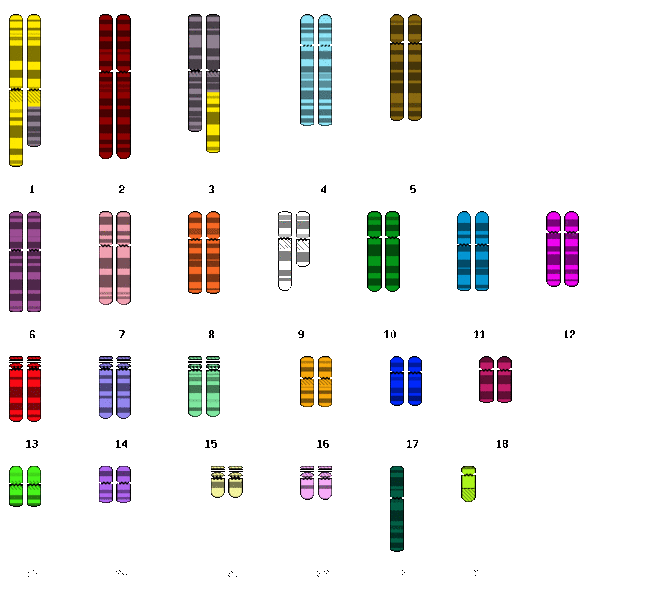
(a) all the different genes found in an ecosystem.

(b) all the different alleles in an interbreeding population.

(c) the alleles in a group of individuals that cause a genetic disease.

(d) the total number of genes found in a species.

Question 2 refers to the Karyotype below.



2. What are the two types of chromosomal mutations seen in the Karyotype above?

(a) Translocation; Addition  
(b) Duplication; Inversion  
(c) Translocation; Deletion  
(d) Duplication; Non-disjunction

3. The genotype of an individual

(a) is always the same as the phenotype.

(b) refers to the visible traits of an individual, what they look like.

(c) refers to the genes a person has for a particular characteristic.

(d) is indicated by the presence of either an X or a Y chromosome.

4. An allele is best defined as

(a) a lethal recessive phenotype..

(b) a lethal dominant phenotype.

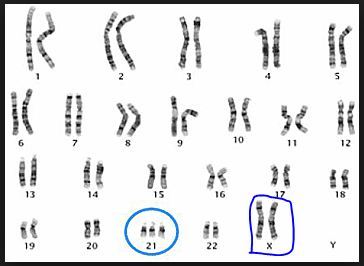
(c) a type of gene only found on one sex chromosome.

(d) an alternative form of a gene at a given locus.

5. For Natural Selection to occur, mutations must be

* 1. germline.
  2. morphologic.
  3. somatic.
  4. autonomic.

Questions 6 and 7 refer to the information and diagram shown below.



Down syndrome is an example of a disorder caused by a mutation. This is known as trisomy-21 and is shown below in the karyotype of an affected female.

6. This type of mutation is a

(a) point mutation.

(b) somatic mutation.

(c) chromosomal mutation.

(d) gene mutation.

7. The cause of this mutation is

(a) non-disjunction.

(b) deletion.

(c) duplication.

(d) translocation.

8.

.

9. Which of the following statements is NOT correct?

(a) Heterozygous individuals with one Tay-Sachs allele appear to have   
 increased resistance to tuberculosis.  
(b) Heterozygous individuals with one sickle-cell anaemia allele have a   
 survival advantage in areas where malaria is prevalent.  
(c) Thalassaemia is an autosomal recessive disease in which anaemia results   
 from the formation of distorted red blood cells.  
(d) Cystic fibrosis is an autosomal recessive disease caused by a mutation   
 preventing the gene from producing a protein that will be able to function   
 in the body.

10.

**PART B: Short Answer Section [Total 25 marks]**

**Question 11 [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 Iraq-Iran War. It found there were 122 different mutated genes in the respiratory pathways of the victims.

(a) The studies described above outline how mustard gas has been shown to cause

mutations. Mustard gas can therefore be classed as a (1 mark)

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(b) Auerbach and Robson were studying chromosomal mutations. Describe **two** types of these mutations that can occur in organisms. (4 marks)

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(c) The 2014 study examined mutated genes. Describe how this type of mutation differs from a chromosomal mutation. (2 marks)

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(d) 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)

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**Question 12 [2 marks]**

West Nile Virus (WNV) is currently on the rise in Western Countries, such as North America, and is known to lethally infect humans. Studies have found that 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 |

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

Patients with WNV: 0.04

Patients without WNV: 0.01

Given that the percentage of **CCR5∆32 homozygotes** in North America is usually 1 percent, compare the frequency of **CCR5∆32 homozygotes** in both populations.

(2 marks)

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**Question 13 [7 marks]**

Anthony Clifford Allison, a South African geneticist, conducted studies on genetic resistance to malaria. This led him to discover the link between malaria and the frequency of the sickle-cell allele in the African population. He found that the majority of Africans with the sickle-cell trait did not develop malaria. The majority without the sickle-cell trait did develop malaria. Malaria can be life threatening so those individuals who had the sickle-cell trait were able to live longer lives than those without the trait.

(a) The sickle-cell trait has an advantage for people living in malaria-prone areas due to a selectively advantageous mutation. Explain what is meant by a selectively advantageous mutation. (3 marks)

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(b) Describe how the link between the sickle-cell allele and malaria can lead to

changes in the allele frequencies in a population. (4 marks)

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**Question 14 [6 marks]**

Using examples, explain how a lethal recessive mutation such as the Tay-Sachs disease can bring about changes in the gene pool through the two mechanisms below:

a) genetic drift (3 marks)

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b) natural selection (3 marks)

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**PART C: Extended Response [Total 20 marks]**

15. For each inherited genetic disease, identify a population in which the disease is prevalent, outline the cause and main symptoms experienced by affected individuals and describe how the disease is inherited and the effect it has on the gene pool of populations.

(a) sickle-cell anaemia (7 marks)

(b) Tay-Sachs disease (6 marks)

(c) thalassemia (7 marks)

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