Logo, company name

Description automatically generated**SOUTHERN RIVER COLLEGE**

**Human Biology**

**Unit 4**

**TASK 7**

**Mutations & Gene Pools Validation (5%)**

**TYPE:** Extended Response

**CONTENT:** Mutations & Gene Pools

You are to complete the following questions, using your knowledge gained from the take home booklet. You are allowed Part A ONLY as notes during this Validation.

Take home booklet /6 marks

References /2 marks

Validation /33 marks

**TOTAL /41 marks**

**\_\_\_\_\_\_%**

*Use the information below to help you answer question 1, 2, 3 and 4.*

It is thought that man evolved in Africa and then migrated from there to populate the world. The table below shows blood group percentages in various ethnic groups. The geographical locations of these populations can be found on the following page, using the map references.

**TABLE 1: Blood group percentages in various ethnic groups**

|  |  |  |  |  |  |
| --- | --- | --- | --- | --- | --- |
|  | **Map reference** | **O (%)** | **A (%)** | **B (%)** | **AB (%)** |
| **Australian Aboriginal** | A | 61 | 39 | 0 | 0 |
| **English** | B | 47 | 42 | 8 | 3 |
| **Eskimos**  **(Alaska)** | C | 38 | 44 | 13 | 5 |
| **Eskimos (Greenland)** | D | 54 | 36 | 23 | 8 |
| **USA (black)** | E | 49 | 27 | 20 | 4 |
| **USA (white)** | F | 45 | 40 | 11 | 4 |
| **Peru (Indians)** | G | 100 | 0 | 0 | 0 |
| **Navajo (Native American Indian)** | H | 73 | 27 | 0 | 0 |



A

E & F

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B

1. Name and outline a factor which explains why Peruvian Indians and Aborigines have evolved with no incidence of B or AB blood groups in their populations. (2 marks)

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1. If the B or AB blood group were to suddenly start to appear in the Aboriginal population, how could this be explained? (2 marks)

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1. Identify and highlight the location of the Eskimo (Alaskan population), Navajo and Peruvian Indians on the map provided.
2. Using the blood group data from Table 1 and the map for the Eskimo (Alaskan population), Navajo and Peruvian Indians, what does this information indicate to us about the spread of people through the Americas’ populations? (3 marks)

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***Use the information below to answer question 5.***

The Andaman and Nicobar islands are located southeast of the **Indian subcontinent**, separated by the **Bay of Bengal** by about 1,300 km. These islands are outlined by the black box.

Table 2 shows the blood group percentages for the populations on these islands (Grand Andamanese and Nicobarese) and those people on the Indian mainland prior to the 2004 earthquake.

**TABLE 2: Blood group percentages for** Grand Andamanese, Nicobarese **and mainland Indian populations** prior to the 2004 earthquake.

|  |  |  |  |  |
| --- | --- | --- | --- | --- |
| **Blood Group** | **O (%)** | **A (%)** | **B (%)** | **AB (%)** |
| **Grand Andamanese (Andaman island)** | 9 | 60 | 23 | 9 |
| **Nicobarese (Nicobar island))** | 74 | 9 | 15 | 1 |
| **Indians (India – General population)** | 37 | 22 | 33 | 7 |

1. In December 2004 a massive earthquake in the Indian Ocean resulted in a tsunami that swept across these islands and coastal India. This may have led to a ‘bottleneck effect” where only a few survive in certain populations.
   1. Explain what resulting effect the bottleneck may have on blood group percentages present in the Grand Andamanese and Nicobarese populations following this natural disaster.   
        
      **Use the data in the table to support your answer**. (6 marks)

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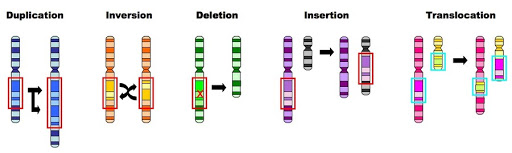
From the information given, natural selection doesn’t appear to be influencing the allele frequencies of the populations.

* 1. Describe how natural selection affects the genotype frequencies of populations in general. (3 marks)

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***Use the diagram below to answer question 6.***



**D**

**E**

**C**

**B**

**A**

1. a) Name the specific type of mutations for each letter below: (2 marks)

B: \_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_

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b) Describe what is occurring in the diagram at A. (1 mark)

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c) Explain the difference between D and E in the diagram above. (2 marks)

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1. For the diseases listed below, describe the effect they have on an individual’s lifespan, the processes that maintain the disease in the population AND why it may be of benefit:
   1. Sickle cell anaemia (3 marks)

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* 1. Tay Sach’s Disease (chronic form) (3 marks)

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1. Describe the difference between alpha and beta thalassemia in terms of the effects these diseases have on the survival of individuals within a gene pool. (6 marks)

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**END OF TEST**

**SOUTHERN RIVER COLLEGE**

**Human Biology**

**Unit 4**

**TASK 7**

**Mutations & Gene Pools MARKING KEY (5%)**

**TYPE:** Extended Response

**CONTENT:** Mutations & Gene Pools

You are to complete the following questions, using your knowledge gained from the take home booklet.

Take home booklet / 6 marks

References / 2 marks

Validation / 33 marks

**TOTAL / 41 marks**

**\_\_\_\_\_\_%**

*Use the information below to help you answer question 1, 2 and 3 and 4.*

It is thought that man evolved in Africa and then migrated from there to populate the world. The table below shows blood group percentages in various ethnic groups.

**TABLE 1:**

|  |  |  |  |  |
| --- | --- | --- | --- | --- |
|  | **O** | **A** | **B** | **AB** |
| **Australian Aboriginal** | 61 | 39 | 0 | 0 |
| **English** | 47 | 42 | 9 | 3 |
| **Eskimos**  **(Alaska)** | 38 | 44 | 13 | 5 |
| **Eskimos (Greenland)** | 54 | 36 | 23 | 8 |
| **USA (black)** | 49 | 27 | 20 | 4 |
| **USA (white)** | 45 | 40 | 11 | 4 |
| **Peru (Indians)** | 100 | 0 | 0 | 0 |
| **Navajo (Native American Indian)** | 73 | 27 | 0 | 0 |

1. Name and outline a factor which explains why Peruvian Indians and Aborigines have evolved with no incidence of B or AB blood groups in their populations. (2 marks)

|  |  |
| --- | --- |
| **Name & Outline** | **Marks** |
| Founder effect | 1 |
| No ‘B’ alleles present in original migrants | 1 |
| **Total** | **2** |

1. If the B or AB blood group were to appear in the Aboriginal population, how could this be explained?

(2 marks)

|  |  |
| --- | --- |
| **Explain** | **Marks** |
| **Migration:** | Any 1 plus matching explanation |
| someone with ‘B’ allele/B or AB blood group has arrived in the population and has passed on through mating |
| **Random mutation:** |
| Either ‘O’ or ‘A’ allele has had a mutation that mimics ‘B’ antigen/allele  OR permanent change which introduces the IB allele into the gene pool which produces the B or AB blood group |
| **Total** | **2** |

1. Identify and highlight the location of the Eskimo (Alaskan population), Navajo and Peruvian Indians on the map provided.

1. Using the blood group data from Table 1 and the map for the Eskimo (Alaskan population), Navajo and Peruvian Indians, what does this information indicate to us about the spread of people through the Americas’ populations? (3 marks)

|  |  |
| --- | --- |
| **Describe** | **Marks** |
| Not a consistent migration | Any 3 |
| probably started in Alaska (all alleles present) |
| moved south in stages |
| Navajo is founder as loss of alleles is gradual |
| Peru has clear founder effect as 2 alleles lost |
| **Total** | **3** |

***Use the information below to answer question 5.***

The Andaman and Nicobar islands are located southeast of the **Indian subcontinent**, separated by the **Bay of Bengal** by about 1,300 km. These islands are outlined by the black box.

Table 2 shows the blood group percentages for the populations on these islands (Grand Andamanese and Nicobarese) and those people on the Indian mainland prior to the 2004 earthquake.

**TABLE 2: Blood group percentages for** Grand Andamanese, Nicobarese **and mainland Indian populations** prior to the 2004 earthquake.

|  |  |  |  |  |
| --- | --- | --- | --- | --- |
| **Blood Group** | **O (%)** | **A (%)** | **B (%)** | **AB (%)** |
| **Grand Andamanese (Andaman island)** | 9 | 60 | 23 | 9 |
| **Nicobarese (Nicobar island))** | 74 | 9 | 15 | 1 |
| **Indians (India – General population)** | 37 | 22 | 33 | 7 |

1. In December 2004 a massive earthquake in the Indian Ocean resulted in a tsunami that swept across these islands and coastal India. This may have led to a ‘bottleneck effect” where only a few survive in certain populations.
2. Explain what resulting effect the bottleneck may have on blood group percentages present in the Grand Andamanese and Nicobarese populations following this natural disaster.   
     
   **Use the data in the table to support your answer**. (6 marks)

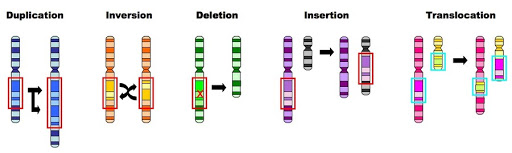
|  |  |
| --- | --- |
| **Describe –** Andamananese at least 1 mark must include data | **Marks** |
| * most likely loss of ‘O’ allele (1 mark) * as only 9% of population carry it (1 mark) | Any 3 |
| * ‘A’ allele will increase further (1 mark) * as 60% carry it (1 mark) |
| * AB blood group could increase (1 mark) * A and B individuals producing offspring of AB blood group (1 mark) |
| **Describe –** Nicobarese at least 1 mark must include data. |  |
| * loss of ‘A’ alleles (1 mark) * as only 9% remain (1 mark) | Any 3 |
| * possibly ‘B’ loss of B allele too (1 mark) * as only 15% (1 mark) * Leading to a loss of AB? (1 mark) |
| * increase in ‘O’ alleles (1 mark) * as already 74% (1 mark) |
| **Total** | **6** |

From the information given, natural selection doesn’t appear to be influencing the allele frequencies of the populations.

1. Describe how natural selection affects the genotype frequencies of populations in general. (3 marks)

|  |  |
| --- | --- |
| **Describe** | **Marks** |
| Individuals with favourable alleles tend to survive and reproduce | Any 3 |
| These traits/characteristics become more abundant in the population |
| Individuals with unfavourable alleles do not survive |
| These traits/characteristics decrease withing a population |
| **Total** | **3** |

*Use the diagram below to answer question 6.*



**D**

**E**

**C**

**B**

**A**

1. a) Name the specific type of mutations for each letter below: (2 marks)

B: Inversion

C: Deletion

b) Describe what is occurring in the diagram at A. (1 mark)

|  |  |
| --- | --- |
| **Describe** | **Marks** |
| a portion of the chromosome is copied/replicated. | 1 |
| **Total** | **1** |

c) Explain the difference between D and E in the diagram above. (2 marks)

|  |  |
| --- | --- |
| **Explain** | **Marks** |
| D: a section of one chromosome is broken off and added to another chromosome. | 1 |
| E: a piece of two chromosomes breaks off and is transferred to the other chromosome (direct swap). | 1 |
| **Total** | **2** |

Must be clear

1. For the diseases listed below, describe the effect they have on an individual’s lifespan, the processes that maintain the disease in the population AND why it may be of benefit:
   1. Sickle cell anaemia (3 marks)

|  |  |
| --- | --- |
| **Describe** | **Marks** |
| Homozygous have reduced lifespan so do not pass on | 1 |
| Heterozygous/carriers are protected against malaria | 1 |
| These individuals therefore survive to pass on the trait | 1 |
| **Total** | **3** |

* 1. Tay Sach’s Disease (chronic form) (3 marks)

|  |  |
| --- | --- |
| **Describe** | **Marks** |
| Homozygous is fatal /reduced lifespan so do not pass on | 1 |
| Genetic drift/barrier the gene flow/inbreeding – Ashkenazi Jews small population isolated by religion/language (explanation of this) | 1 |
| Protection against tuberculosis if Heterozygous/carrier | 1 |
| **Total** | **3** |

1. Describe the difference between alpha and beta thalassemia in terms of the effects these diseases have on the survival of individuals within a gene pool. (6 marks)

|  |  |
| --- | --- |
| **Describe - Alpha** | **Marks** |
| Mild, moderate or severe symptoms depening on the number of genes the individual inherits (up to 4 genes) | 1 |
| Fatal if individuals have all 4 genes effected/ Fatal if homozygous for condition | 1 |
| Heterozygous provides resistance against malaria | 1 |
| **Describe - Beta** | 1 |
| Homozygous is severe but not lethal/fatal | 1 |
| Does not offer resistance against malaria | 1 |
| Gene pool isolated due to cultural and grographical ie. marriage between cousins along the Mediteranean sea (founder effect also in play here) | 1 |
| **Total** | **6** |

|  |  |
| --- | --- |
| **Take home booklet** | **Marks** |
| Data in tables attempted (minor errors) | 1 |
| All data in tables correct (see answers below, correct to 1 dp) | 1 |
| Other questions completed – basic (ie. Writing in every box) | 1 |
| Other questions completed – in detail | 1 |
| Completed A3 sheet – basic | 1 |
| Completed A3 sheet – in detail | 1 |
| **References** |  |
| 3 references in *attempted* APA 6th edition format. | 1 |
| At least 3 plus references in correct APA 6th edition format. | 1 |
| **Total** | **8** |

**Take home part answers:**

1. Record the following in the tables provided:

a) Count the allele frequencies in both populations.

b) Count the number of allele combinations in both populations.

c) Calculate the allele frequencies as percentages for both, round to 1 decimal place.

|  |  |  |  |  |  |  |  |  |
| --- | --- | --- | --- | --- | --- | --- | --- | --- |
| **Original population** | | **Number counted** | **%** |  | **Island population** | | **Number counted** | **%** |
| **Allele types /84** | IA | 33 | 39.3 |  | **Allele types**  **/34** | IA | 16 | 47.1 |
| IB | 10 | 11.9 |  | IB | 0 | 0 |
| i | 41 | 48.8 |  | i | 18 | 52.9 |
| **Allele combinations /42** | IAIA | 9 | 21.4 |  | **Allele combinations**  **/17** | IAIA | 3 | 17.7 |
| IAi | 10 | 23.8 |  | IAi | 10 | 58.8 |
| IBIB | 0 | 0 |  | IBIB | 0 | 0 |
| IBi | 5 | 11.9 |  | IBi | 0 | 0 |
| IAIB | 5 | 11.9 |  | IAIB | 0 | 0 |
| ii | 13 | 31.0 |  | ii | 4 | 23.5 |

|  |  |  |  |  |  |  |  |  |
| --- | --- | --- | --- | --- | --- | --- | --- | --- |
| **Original phenotype frequencies** | | | |  | **Island phenotype frequencies** | | | |
| **Type A** | **Type B** | **Type AB** | **Type O** |  | **Type A** | **Type B** | **Type AB** | **Type O** |
| 45.2 | 11.9 | 11.9 | 31.0 |  | 76.5 | 0 | 0 | 23.5 |

2. Use the following formula to calculate a gene (allele) frequency:

**Gene (allele) frequency =**

**number of specific alleles ÷ total number of alleles x 100**

*NOTE: round to 1 decimal place*