***Human Biology ATAR – Task 11: Science Inquiry***

***The genetics of sickle anaemia (5%)***

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| Name: | | | |
| Time allowed: 1 Lesson | | | |
| **Section** | Your Mark | Marks available | Percentage of Investigation |
| Research questions |  | 20 | 100% |

**Declaration of Authenticity**

I (Student Name) \_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_ declare that this work is my own and I have not plagiarised from any source.

Signature:  
  
Date:

***Sickle cell AND Inheritance - Information***

In 1910 an American physician, J.B. Herrick, examined the blood of an African American boy with a mysterious disease. The red blood cells of the boy were shaped like crescents, or sickle shaped. Soon after, Herrick wrote an account of his discovery, other physicians uncovered cases of the same illness. It proved to be quite common amongst people of WestAfricandescent.

In the disease, now called Sickle-Cell Anaemia, newly formed red blood cells are normal in shape; but when the oxygen in their haemoglobin is released to the body tissues, most of them change to the abnormal sickle shape.

The sickle cells are destroyed in the spleen. This reduction in the number of blood cells cuts down the amount of oxygen available for the body cells. Also, because the sickle cells are much less flexible than normal red blood cells, they do not pass through capillaries as easily. By clogging up the capillaries, sickle cells further reduce the efficiency of the circulation. People with Sickle-cell anaemia usually die in childhood.

Individualswith less severe cases of ‘sickling’ may produce sickled red blood cells when the supply of oxygen is low [as at high altitudes] or when their need for oxygen increases [as during strenuous physical exercise]. Such persons are said to have the ‘Sickle-cell trait’ (also called Sickle-cell disease). Hence, they are considered to be heterozygous. So they have some normal red blood cells and some sickle shaped red blood cells.

It has been shown that sickling is a characteristic of the haemoglobin in the cells. In cells that sickle, the haemoglobin molecule differs from a normal molecule at just 2 points along the chains of 574 amino acids which make up the protein part of the molecule.

***Write your answers on the answer sheet provided***

**The genotypes are below.**

* ***HbS = Sickle-cell gene***
* ***HbA = normal gene***

1. Write down the genotype of a person with (2 marks)
   1. Sickle-cell anaemia\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_
   2. Sickle-cell trait\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_

*The gene that brings about the formation of sickle cell haemoglobin is rare in most human populations. In some parts of Africa, however, the sickle cell trait is found in as much as 40 per cent of the population*.

1. When a heterozygous individual mates with another heterozygous individual, what is the percentage of the offspring that would be expected to be homozygous for the Sickle-cell Anaemia? Show all working (punnet square) (3 marks)

1. What effect would you expect the death of children with Sickle-cell anaemia to have on the frequency of the gene for sickling in any population? (1 mark)

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1. Which would you expect to leave more children surviving into adulthood - couples mating who both have Sickle-cell trait *(heterozygous individuals)* or couples mating who both have normal red blood cells? Why? Show evidence to support your argument (3 marks)

*There is no evidence that the frequency of the gene for sickling is becoming less in African populations. Therefore a biological problem arises: how can the gene for sickling be so great when natural selection works so strongly against the gene? Biologists have developed at least three hypotheses to account for the high frequency of the sickling gene in African populations. One involves mutation rates, a second involves fertility and a third involves resistance to disease.*

*Through genetic reasoning and mathematical techniques, it is possible to calculate the rate at which genes are lost from the population gene pool by natural selection. This rate has been found to be 100 times the average rate of mutation at any one place on a gene in human chromosomes.*

1. Does this finding support or not support the *mutation rates* hypothesis? Explain.

(2 marks)

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1. Does this finding support or not support the *fertility* hypothesis? Explain (2 marks)

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*As data on sickling was collected, the frequencies of the Sickle-cell trait in various populations were plotted on maps. It became clear that the gene is most common in a belt extending across Central and West Africa. In the same region, malaria and hookworm are common diseases. Malarial parasites multiply inside red blood cells; hookworms feed on the lining of the intestines.*

1. Which of the two diseases would be most likely to be associated with sickling? Explain. (2 marks)

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*The present frequency of Sickle-cell trait in parts of Africa from which the ancestors of African Americans came indicates that the early African American population contained about 22 per cent sicklers.*

1. How would mixing with the European population have caused this frequency to change? Use a punnet square to demonstrate how this happens (3 marks)

*In the United States of America, control measures have almost eliminated the transmitter of Malaria - the Anopheles mosquito.*

1. Recalling that an individual homozygous for sickling usually dies before reaching reproductive age, what would you expect to happen to the frequency of the gene for sickling in the United States. Why? (2 marks)

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