**ATAR HUMAN BIOLOGY**

**UNIT 2**

**Task 8 – Science Inquiry: DNA**

NAME: \_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_

DUE DATE: \_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_ WEIGHTING: 5 %

TEACHER: \_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_ MARK: \_\_\_ / 35 = \_\_\_\_\_\_\_ %

**Conditions**

Time for the task:

* Part 1: 5 minutes reading time
* Part 2: Carry out a practical investigation into epigenetics and DNA, process results. (30 minutes)
* Part 3: 1 hour 25minutes for in-class practical write up and validation questions

**Introduction**

In the field of epigenetics, scientists study how chemical tags attach themselves to DNA or the structures surrounding the DNA. These chemical tags can control whether a certain gene will be transcribed and translated into it’s corresponding protein. Because these chemical tags are independent of the DNA sequence itself, they are considered to be epigenetic factors.

These chemical tags can modify the chromatin, but their effects can either remain in place or can be altered throughout a person’s life. The addition of an acetyl group (acetylation) to DNA affects the histones and enhances transcription; in contrast the addition of a methyl group (methylation) to some regulatory regions of the DNA can reduce transcription as well as modify histones. These modifications, together with other regulatory mechanisms, are important during development as they are responsible for determining what an embryonic stem cell becomes (differentiation) and also ensuring it continues to be the specific cell it’s differentiated into.

Epigenetic modifications can also occur in response to the environment one lives in and their diet. How diet affects epigenetics is not fully understood, but many connections have been made between diet and human conditions. For example during the winter of 1944-1945, the Netherlands suffered a terrible famine as a result of the German occupation and the population’s nutritional intake dropped to fewer than 1000 calories per day. Women continued to conceive and give birth during these times, but the babies were very small. These babies, now adults have shown they have a higher rate of chronic conditions such as diabetes, cardiovascular disease and obesity than their siblings showing something that occurred before you were born can influence your life up to some 60 years later. These babies went on to have children of their own which were also small by comparison to the rest of the population not affected by the famine. It was discovered that those exposed to famine whilst in the womb have a lower degree of methylation of some genes (eg. growth factor II gene). This study has shown that epigenetic factors can be passed down through generations where once it was thought they were reversible, confirming there is a major connection between epigenetic factors and diet.

The links between diet and epigenetics are far from clear but the Dutch mothers showed that when certain nutrients are missing from the diet that changes occur to the epigenetic markers on our DNA. One of the most important epigenetic changes is methylation which usually labels genes that are switched off. In order to maintain the pattern of methylation through cell division, new methyl groups are stuck on to freshly-copied DNA. This requires a constant supply of new methyl groups, which can come directly from our food or can be made from the chemicals we eat like folic acid. In addition acetyl groups are also important in a diet to ensure genes can also be switched on. Recently links have been established between the lack of methylation of the DNA and the development of cancer. This indicates that diets containing the building blocks for methylation in the body may enhance your chances of not developing cancer. These types of foods include folic acid, leafy vegetables, peas, beans, sunflower seeds, garlic and liver. Other foods such as turmeric, green tea and soya have been shown to decrease DNA methylation.

Epigenetic changes have been linked to many diseases. As these changes can be reversible, there is great interest in finding molecules, especially those in diet, that might undo these damaging changes and prevent the further development of such conditions. A diet rich in foods that maintain these important methylation patterns and low in substances that remove them, would seem the best way to maintain one’s health.

**Purposes of the investigation:**

* To explore the concept of epigenetics to explain apparently inherited conditions
* To examine factors that affect gene expression
* To understand the influence of the environment on gene expression.

**Hypothesis:**

If you increase the amount of foods that promote DNA methylation in your diet then the more chemical tags will attach to the DNA, causing more gene methylation.

**Materials**

50cm piece of string

Small balls of plastercine, flattened, fixed round the string to represent the genes on the DNA (string)

About 50 chads (circles of paper produced from a hole punch (represent the amount of leafy vegetables in the diet)

**Procedure:**

1. Attach the plastercine balls along the length of the string – choose even or random spacing. Fold the plastercine around the string and then flatten to a small disc shape approximately 5mm diameter, on the string as shown below.
2. To stimulate the effect of increasing leafy vegetables in the diet on methylation:
3. Arrange the string chromosome into any shape on a flat surface, it must contain no coils.
4. Take 10 chads and drop them from 30cm above the string chromosome.
5. The genes (labels) that in contact with a chad have been methylated. Note down how many genes were methylated. Remove all the chads
6. Repeat steps I – iii two more times and record your results in a table in the space below.
7. Repeat steps I – iv, but increase the numbers of chads (leafy vegetables in the diet) by 10 each time until 50 chads has been reached.
8. Process your results and show them in a table. (2 marks)
9. Then present them in a graphical form. **Include** an explanation as to why you choose to display (graph) the data the way you did. (3 marks)

**Analysis and evaluation:**

a) Describe the trend and pattern(s) in your data. (2 marks)

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b) State how your data relates to your hypothesis. (2 marks)

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c) Use your knowledge and understanding of epigenetic factors to explain your results. (3 marks)

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d) Discuss the validity of the investigation and make a relevant suggestion as to how the validity of the experiment could be improved. (4 marks)

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**Validation Questions**

1. What problems would you envisage a person could experience if their diet consisted solely of leafy vegetables? (2 marks)

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2. In terms of a person’s epigenome, why is it important that person maintains a healthy diet of food containing both methyl and acetyl groups. (2 marks)

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3. The changes produced by methylation are localised to the particular cells in which it occurs. If for example the melanin producing gene was silenced in a germ cell of the dermis of the skin, it would show as a white patch on the skin. If it occurred in the epidermal cell that is about to be removed by natural sloughing of the skin cells, then there would be no observable effect.

Methylation is affected by the following:

* Diet
* Stress
* Heavy metals
* Pesticides
* Diesel exhaust fumes
* Tobacco smoke

Use your understanding of epigenetics to explain the following situations:

1. Some smokers develop lung cancer after smoking cigarettes for a very short time, while some long-term smokers never develop lung cancer. (2 marks)

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1. More cancers occur in older people. (2 marks)

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4. In humans the FOXP3 gene is associated with the functioning of the immune system. When a methyl group is added to the cytosine bases in CpG islands in the regulating region of this gene, gene expression is switched off resulting in T-cell function being impaired. In people with methylation of the FOXP3 gene their immune system function does not function as it should and asthma can develop.

a) Using identical twins, explain why one twin can develop asthma whilst the other could have normal immune system functioning and not develop asthma. (2 marks)

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b) How does the data from your experiment support the answer you gave in part a)? (1 mark)

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5. All mammals have the agouti gene. Studies of this gene in mice have shown that when the gene is completely unmethylated mice coat colour is yellow; they are obese and prone to diabetes and cancer. When the gene is methylated (normal mice) the mice coat colour is brown and they have a low risk of disease. Both the fat yellow mice and the healthy brown mice are genetically identical.

a) Explain what would happen if a pregnant yellow mouse was fed a methyl rich diet? (3 marks)

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b) Chemicals can also effect the epigenome. Bisphenol A (BPA) is used to make plastic water bottles and has been shown to reduce methylation of the agouti gene. Scientists have studied the effect of feeding BPA to yellow coated pregnant mice. After giving birth, studies have revealed that the pregnant mouse gave birth to brown coated babies. Explain how this could occur. (2 marks)

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6. Studies of identical twins have shown that during their life they can develop genetic conditions such as short sightedness, obesity and cancer. The development of these types of conditions has been linked to lifestyle and diet and can be reversed over time. In comparison the babies born to the Dutch mothers during the famine continue to have small babies and be more susceptible to certain disease regardless of their life style and diet. Using your knowledge of epigenetic factors, discuss the differences between these two groups of people.

(3 marks)

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