|  |  |  |
| --- | --- | --- |
| **Data, analysis & conclusion** |  |  |
| Data processing | /2 | 1- raw data organised, presented in a table and some processing/averaging  2- raw data organised in a table with headings – averages calculated, any outliers identified |
| Graphing | /3 | * Graph drawn, one mistake (1) * Perfect graph, all five points correct (both marks) * Explanation of why choose the graph (1) – need reference to continuous data so line graph |
| Analysis and Evaluation | /7 | Trends and patterns in the data are described. (total = 2)   * The greater the number of chads dropped the more genes methylated (1) * Comment on if the relationship is linear/random whatever is appropriate to data. (1)   Hypothesis (total of 2)   * The more leafy vegetables you eat the greater the chance that the genes will be methylated (1 – link to results), * which supports the hypothesis that if you eat more foods that contain chemicals that promote methylation then more gene methylation will occur. (1 – link to hypothesis)   Science knowledge and understanding is used to explain the results. (total = 3)   * diet is a factor that can affect your genome determining which genes can be expressed or switched off * The more chemicals (methyl groups) that promote methylation in your diet the more chance they will cause more methylation of genes * Optimal amount of chemicals in the body/cells means all genes that need to be methylated can be * and can be passed on when the cell replicates |
| Validity | /4 | 2 marks – a statement about experiment and then linked to whether that makes the experiment valid or not:   * In a real life situation a person would need to have a balanced diet of both methylating and acetylating chemicals to stay healthy/ need to eat other things in diet to ensure that nutrients are absorbed and can be used/other factors also affect whether a gene can be methylated or not, not just adequate vegetables in diet   1 mark - the experiment does not test this so it is not totally valid, must relate back to science and explain why not valid in scientific terms  1 mark – valid suggestion on how to improve the experiment |

**Validation Questions**

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

***Too many genes would become methylated when they shouldn’t be***

***Meaning genes switched off when they need to be switched on/prevents acetylation of genes/hard for genes to be demethylated***

***Malnutrition – leafy vegetables do not provide a balance of nutrients(strictly speaking answers the question)***

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)

***To remain healthy a person relies on genes being switched on and off***

***Sometimes genes that are off need to be switched on so you need both chemicals available to get the balance right***

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
* 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 long-term smokers never develop lung cancer. (2 marks)

***- tobacco smoke has not affected the cells/DNA to cause cancer in long term smokers***

***- methylation is a random event, even though it is increased by smoking.***

***- balanced out by other factors in the diet***

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

***- have had greater exposure to sources of methylation in the environment over their lifetime than younger people.***

***- cumulative effects /events***

***- more time for reduction in epigenetics factors that prevent the onset***

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) Explain why, with identical twins, one twin can develop asthma whilst the other could have normal immune system functioning so no asthma develops.

(2 marks)

***DNA identical, epigenome different***

***One child exposed to environment that caused increased methylation, the other is not***

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

***Link to experiment – more exposure to environmental factor, more chance the gene will be methylated same as experiment***

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 feed a methyl rich diet? (3 marks)

***most babies will be brown***

***healthy for life***

***chemical tags put in place whilst in the womb remain throughout life***

b) Chemicals can also affect 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, studied have revealed that the pregnant mouse gave birth to brown coated babies. Explain how this could occur. (2 marks)

***something else they ate in their diet counteracted the reduction in methylation***

***therefore the methyl tags could attach because a substance in the diet helped them /or prevented acetylation of the gene***

***random -may not be enough chemical to prevent methylation of all genes, agouti gene spared***

6. Studies of identical twins have shown that during their life they can develop and reverse different genetic conditions such as short sightedness, obesity and cancer. The development of these types of conditions has been linked to life style and diet. In comparison the babies born to the Dutch mothers during the famine continue to have small babies and be more susceptible to certain diseases regardless of their life style and diet. Using your knowledge of epigenetic factors discuss the differences between these two groups of people. (3 marks)

* ***chemical tags put in place when baby develops in the womb remain in place throughout life and can be passed down through generations / permanent***
* ***identical twins have identical DNA so methylation/acetylation must be responsible for genetic changes – epigenetic factors/environment***
* ***chemical tags that are put in place once the baby has developed and being born can be altered/changed / not permanent***