# Gene Expression and Cancer: AQA A Level Biology Exam Questions by T

14 questions · 42 marks · about 55 minutes. Answer in the spaces, then check the mark schemes at the end.

**Question 1 · A level Paper 2 · June 2018 · Q02.2 · 3 marks**

**0 2 . 2** Describe how alterations to tumour suppressor genes can lead to the development of tumours.

**[3 marks]**


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**Question 2 · A level Paper 2 · June 2022 · Q10.1 · 3 marks**

**1 0** Read the following passage.

*BRCA1* and *BRCA2* are human genes that code for tumour suppressor proteins. Mutations in *BRCA1* and *BRCA2* can cause cancer. Specific inherited mutations in these genes increase the risk of female breast cancers and ovarian cancers and have been associated with increased risks of several other types of cancer. Genetic testing, using DNA from saliva, can screen for all known harmful mutations in both genes.

ER-positive breast cancers have receptors for the hormone oestrogen. These cancers develop as a result of increased oestrogen concentrations in the blood. Effective treatment of ER-positive breast cancers often involves the use of drugs which have a similar structure to oestrogen.

Blood tests can be used to test for cancers. Men with prostate cancer have a high concentration of prostate-specific antigen (PSA) in their blood. Urinary infections and a naturally enlarged prostate can also increase concentrations of PSA.

Recent research has indicated that several cancers result from epigenetic abnormalities. Treatment with drugs might be able to reverse the epigenetic changes that cause cancers.

Use the information in the passage and your own knowledge to answer the following questions.

**1 0 . 1** *BRCA1* and *BRCA2* are human genes that code for tumour suppressor proteins. Mutations in *BRCA1* and *BRCA2* can cause cancer (lines 1–2). Explain how.

**[3 marks]**


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**Question 3 · A level Paper 2 · June 2023 · Q08.1 · 3 marks**

**0 8 . 1** The *KRAS* gene codes for a protein called K-Ras. The protein relays signals from outside a cell to a cell's nucleus, stimulating cell division. An alteration in the *KRAS* gene produces an oncogene which can cause a tumour to develop.

Suggest and explain how an alteration in the *KRAS* gene can cause a tumour to develop.

**[3 marks]**


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**Question 4 · A level Paper 3 · June 2017 · Q05.4 · 3 marks**

**0 5 . 4** Sometimes, a mutagenic agent causes DNA to break. A different enzyme called ATM binds to the broken DNA. This leads to the activation of a protein coded for by a tumour suppressor gene. The effect of ATM binding is to stop cell division until DNA is repaired.

A mutation could result in a person having non-functional forms of the gene that produces ATM.

What can you predict about the possible effects of having a non-functional form of ATM?

**[3 marks]**


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**Question 5 · A level Paper 3 · November 2020 · Q05.2 · 3 marks**

Increased methylation of the promoter region of a tumour suppressor gene causes one type of human throat cancer.

In this type of throat cancer, cancer cells are able to pass on the increased methylation to daughter cells. The methylation is caused by an enzyme called DNMT.

Scientists have found that a chemical in green tea, called EGCG, is a competitive inhibitor of DNMT. EGCG enables daughter cells to produce messenger RNA (mRNA) from the tumour suppressor gene.

**0 5 . 2** Suggest how EGCG allows the production of mRNA in daughter cells.

**[3 marks]**

**Question 6 · information from earlier in the question**

**0 6** Myelodysplastic syndromes (MDS) are a group of malignant cancers. In MDS, the bone marrow does not produce healthy blood cells.

Haematopoietic stem cell transplantation (HSCT) is one treatment for MDS. In HSCT, the patient receives stem cells from the bone marrow of a person who does not have MDS. Before the treatment starts, the patient's faulty bone marrow is destroyed.


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**Question 6 · A level Paper 2 · November 2021 · Q06.2 · 3 marks**

**0 6 . 2** MDS can develop from epigenetic changes to tumour suppressor genes. In some patients, the drug AZA has reduced the effects of MDS. AZA is an inhibitor of DNA methyltransferases. These enzymes add methyl groups to cytosine bases.

Suggest and explain how AZA can reduce the effects of MDS in some patients.

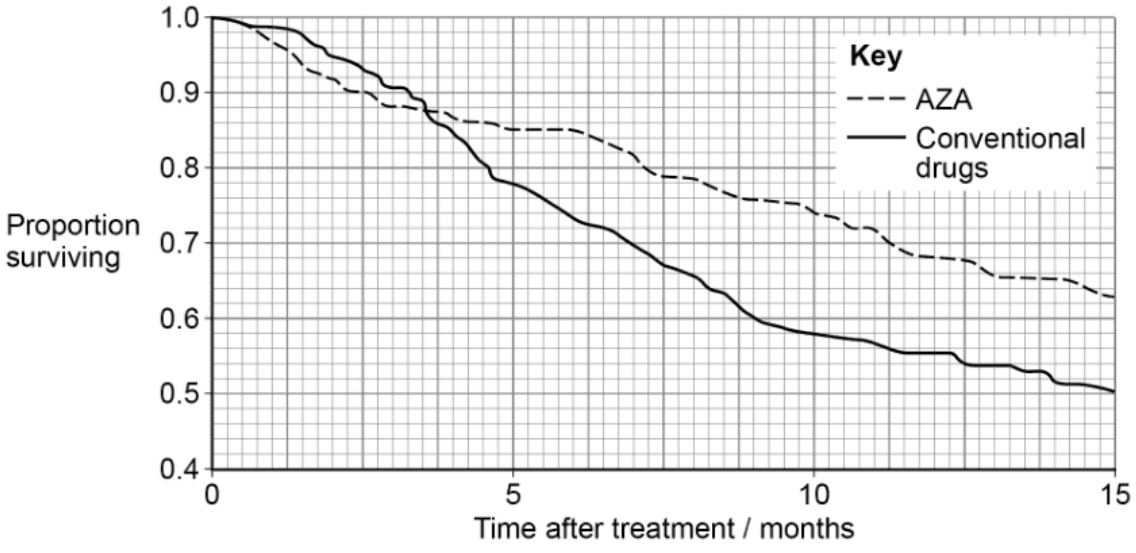
**[3 marks]**

**Question 7 · information from earlier in the question**

Scientists investigated the effectiveness of AZA in patients with MDS.
A total of 360 patients were randomised in the ratio of 1:1 to receive AZA or conventional drugs (control).

**Figure 6** shows the scientists' results.

**Figure 6**





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**Question 7 · A level Paper 2 · November 2021 · Q06.4 · 2 marks**

**0 6 . 4** Use **Figure 6** and the information provided to calculate the difference in the number of patients surviving at 10 months after treatment with AZA compared with conventional drugs.

**[2 marks]**

Answer _____

**Question 8 · information from earlier in the question**

**1 0** Read the following passage.

*BRCA1* and *BRCA2* are human genes that code for tumour suppressor proteins. Mutations in *BRCA1* and *BRCA2* can cause cancer. Specific inherited mutations in these genes increase the risk of female breast cancers and ovarian cancers and have been associated with increased risks of several other types of cancer. Genetic testing, using DNA from saliva, can screen for all known harmful mutations in both genes.

ER-positive breast cancers have receptors for the hormone oestrogen. These cancers develop as a result of increased oestrogen concentrations in the blood. Effective treatment of ER-positive breast cancers often involves the use of drugs which have a similar structure to oestrogen.

Blood tests can be used to test for cancers. Men with prostate cancer have a high concentration of prostate-specific antigen (PSA) in their blood. Urinary infections and a naturally enlarged prostate can also increase concentrations of PSA.

Recent research has indicated that several cancers result from epigenetic abnormalities. Treatment with drugs might be able to reverse the epigenetic changes that cause cancers.

Use the information in the passage and your own knowledge to answer the following questions.


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**Question 8 · A level Paper 2 · June 2022 · Q10.3 · 3 marks**

**1 0 . 3** Effective treatment of ER-positive breast cancers often involves the use of drugs which have a similar structure to oestrogen (lines 9–10).

Suggest and explain how these drugs are an effective treatment of ER-positive breast cancers.

**[3 marks]**

**Question 9 · A level Paper 2 · June 2018 · Q02.3 · 2 marks**

**0 2 . 3** A type of malignant tumour cell divides every 8 hours.

Starting with one of these cells, how many tumour cells will be present after 4 weeks? Assume none of these cells will die.

Give your answer in standard form.

**[2 marks]**

Answer = _____


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**Question 10 · information from earlier in the question**

**1 0** Read the following passage.

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ER-positive breast cancers have receptors for the hormone oestrogen. These cancers develop as a result of increased oestrogen concentrations in the blood. Effective treatment of ER-positive breast cancers often involves the use of drugs which have a similar structure to oestrogen.

Blood tests can be used to test for cancers. Men with prostate cancer have a high concentration of prostate-specific antigen (PSA) in their blood. Urinary infections and a naturally enlarged prostate can also increase concentrations of PSA.

Recent research has indicated that several cancers result from epigenetic abnormalities. Treatment with drugs might be able to reverse the epigenetic changes that cause cancers.

Use the information in the passage and your own knowledge to answer the following questions.

**Question 10 · A level Paper 2 · June 2022 · Q10.4 · 2 marks**

**1 0 . 4** Blood tests can be used to test for cancers (line 11). However, the results of blood tests may **not** be conclusive when testing for prostate cancer. Explain why.

**[2 marks]**


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**0 8 . 2** Alterations in the *KRAS* gene can cause colorectal cancer (CRC). Scientists investigated the survival time of the following three groups of CRC patients who had type II diabetes.

Group **A** – received no drug to treat type II diabetes
Group **B** – received the drug metformin to treat type II diabetes
Group **C** – received a combination of drugs (including metformin) to treat type II diabetes

The scientists used a statistical test to compare the survival time of these patients with CRC patients with no history of type II diabetes.

**Table 1** shows some of the results obtained by the scientists.

**Table 1**

| Group | Mean survival time of CRC patients / months | Probability value (P) |
|---|---|---|
| No history of type II diabetes | 32.2 | – |
| A | 21.3 | 0.007 |
| B | 49.7 | 0.022 |
| C | 38.3 | 0.636 |

Using the information provided, what can you conclude about the effects of type II diabetes and the different drug treatments on the survival time of CRC patients?

**[5 marks]**


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Scientists investigated the relationship between one type of variable number tandem repeat (VNTR) and the risk of breast cancer occurring in women.

The scientists determined the risk of breast cancer occurring in women:

- with 2, 3 or 4 repeats of the VNTR
- **without** the VNTR.

They calculated the odds ratio (OR) of breast cancer occurring with 2, 3 or 4 repeats of the VNTR.

An OR is a measure of how strongly an outcome is associated with a risk factor.

The OR formula is:

risk of breast cancer occurring **with** the VNTR : risk of breast cancer occurring **without** the VNTR

**Table 5** shows the scientists' results.

**Table 5**

| Number of VNTR repeats | Odds ratio |
|---|---|
| 2 | 0.55 : 1 |
| 3 | 1.58 : 1 |
| 4 | 1.94 : 1 |

**0 8 . 4** A student concluded that the number of repeats of the VNTR is associated with the risk of breast cancer occurring in women.

Evaluate the student's conclusion.

**[4 marks]**


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**Question 13 · information from earlier in the question**

**0 2** Testosterone is a steroid hormone that belongs to a group of male sex hormones called androgens.

**Question 13 · A level Paper 2 · November 2020 · Q02.4 · 3 marks**

The gene that codes for the AR has a variable number of CAG repeats.
Some studies have shown an association between the number of CAG repeats and the risk of developing prostate cancer.

**Table 1** shows the results of a statistical test from one study.

**Table 1**

| Number of CAG repeats in the *AR* gene | Probability (P) value |
|---|---|
| ⩽ 16 | 0.02 |
| ⩽ 17 | 0.30 |
| ⩽ 18 | 0.07 |
| ⩽ 19 | 0.09 |
| ⩾ 20 | 0.06 |

**0 2 . 4** What can you conclude from the data in **Table 1**?

**[3 marks]**


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**Question 14 · information from earlier in the question**

Increased methylation of the promoter region of a tumour suppressor gene causes one type of human throat cancer.

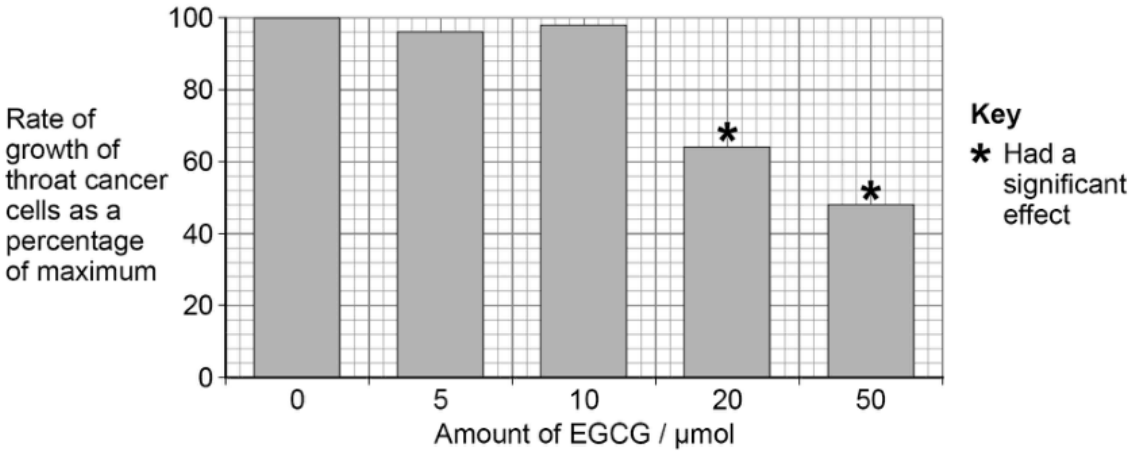
In this type of throat cancer, cancer cells are able to pass on the increased methylation to daughter cells. The methylation is caused by an enzyme called DNMT.

Scientists have found that a chemical in green tea, called EGCG, is a competitive inhibitor of DNMT. EGCG enables daughter cells to produce messenger RNA (mRNA) from the tumour suppressor gene.


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The scientists investigated the effect of different amounts of EGCG on the growth rate of the throat cancer cells grown *in vitro*. Their results are shown in **Figure 2**.

**Figure 2**




**0 5 . 3** A reporter who reviewed all of this work concluded that drinking green tea could be a cure for cancer.

Suggest **three** reasons why his conclusion might **not** be valid.

**[3 marks]**

1 _____

2 _____

3 _____


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## Mark schemes

**Question 1 · A level Paper 2 · June 2018 · Q02.2 · 3 marks**

| | | |
|---|---|---|
| 02.2 | 1.(Increased) methylation (of tumour suppressor genes);<br>2.Mutation (in tumour suppressor genes);<br>3. Tumour suppressor genes are not transcribed/expressed<br>**OR**<br>Amino acid sequence/primary structure altered;<br>4. (Results in) rapid/uncontrollable cell division; | 3 max |

1. Accept abnormal methylation or hypermethylation

1. Ignore decreased acetylation of histones

3. Accept mRNA for transcription/transcribed

3. Accept tertiary structure altered

3 Accept different amino acid

3. Ignore reference to protein not being formed

4. Accept cell division cannot be regulated

4 Ignore growth


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**Question 2 · A level Paper 2 · June 2022 · Q10.1 · 3 marks**

| | | |
|---|---|---|
| 10.1 | 1. Change in <u>DNA</u> base sequence/triplet;<br>2. Change in (sequence of) <u>amino acids</u><br>**OR**<br>Change in primary/tertiary/$3^0$ structure;<br>3. (Results in) rapid/uncontrollable cell division; | 3<br>(3 x AO2) |

| |
|---|
| 2. Ignore reference to protein not being formed. |
| 2. Reject (different) amino acids formed. |
| 2. Ignore 3D structure. |
| 3. Accept cell division cannot be regulated. |
| 3. Ignore growth. |
| 3. Accept cell replication but ignore cell reproduction. |


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**Question 3 · A level Paper 2 · June 2023 · Q08.1 · 3 marks**

| | | |
|---|---|---|
| **08.1** | 1. Mutation (in *KRAS* gene)<br>**OR**<br>Change in <u>base</u> sequence (of *KRAS* gene);<br>2. Change in (signalling) protein<br>**OR**<br>More (signalling) protein/K-Ras produced;<br>3. (Results in) rapid/uncontrollable cell division; | 3<br>(3 x AO2) |

1. Accept named mutation e.g. substitution.

1. Accept mutation in promoter gene.

1. Ignore epigenetic modifications.

2. Accept change in amino acid sequence (of protein).

2. Reject 'no protein produced' or suggests a non-functional protein is produced.

3. Accept cell division cannot be regulated.

3. Ignore growth.

3. Reject meiosis for cell division, but accept mitosis


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**Question 4 · A level Paper 3 · June 2017 · Q05.4 · 3 marks**

| | | |
|---|---|---|
| 05.4 | 1. ATM will not bind to (broken) DNA;<br>2. DNA not repaired/ cell still has broken DNA;<br>3. Cell division continues/tumour forms;<br>4. Tumour suppressor (gene) not effective/ not activated;<br>5. May have no effect in diploid/heterozygous (organism);<br>6. (Which) still has a functional ATM/ATM gene; | 3 max |

**Question 5 · A level Paper 3 · November 2020 · Q05.2 · 3 marks**

| | | |
|---|---|---|
| **05.2** | 1. (EGCG) binds to <u>active site</u> of DNMT;<br>2. (DNMT) cannot methylate (promoter region of tumour suppressor gene);<br>3. Transcription(al) factor(s) can bind (to promoter region);<br>4. RNA polymerase (stimulated/activated); | 3 max |

1. Ignore active site changes shape
1. Ignore 'forms enzyme-substrate' complex
2. Accept less methylation (of promoter region/tumour suppressor gene)


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**Question 6 · A level Paper 2 · November 2021 · Q06.2 · 3 marks**

| | | |
|---|---|---|
| **06.2** | 1. (AZA) reduces methylation (of DNA/cytosine/gene);<br>2. (Tumour suppressor) gene is transcribed/expressed;<br>3. Prevents rapid/uncontrollable cell division<br>**OR**<br>Cell division can be controlled/stopped/slowed; | 3 |

1. Reject any reference to mutation.

2. Accept mRNA produced for transcription/transcribed.

2. Ignore gene is 'switched on' or activated but allow protein is formed.

3. Ignore growth.

**Question 7 · A level Paper 2 · November 2021 · Q06.4 · 2 marks**

| | | |
|---|---|---|
| **06.4** | 1. Correct answer of 29/28.8 = **2 marks**;;<br>2. Working shows 0.74 **and** 0.58 = **1 mark**<br>**OR**<br>58/57.6 = **1 mark**<br>**OR**<br>28 = **1 mark**; | 2 |


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**Question 8 · A level Paper 2 · June 2022 · Q10.3 · 3 marks**

| | | | |
|---|---|---|---|
| 10.3 | 1. (Drug) binds to (oestrogen/ER) receptor;<br>2. Prevents binding of oestrogen/hormone;<br>3. No/fewer transcription factor(s) bind to promoter<br>**OR**<br>RNA polymerase not stimulated/activated; | 3<br>(3 x AO2) | 1. Accept (inactive) transcription factor for receptor.<br>1 and 2. Reject active site/enzyme - substrate complex once only. |

**Question 9 · A level Paper 2 · June 2018 · Q02.3 · 2 marks**

| | | | |
|---|---|---|---|
| 02.3 | 1. Correct answer of 1.9/1.93 x $10^{25}$ = **2 marks**;;<br>2. Incorrect answer but shows 84 = **1 mark**<br>**OR**<br>28 x 3 = **1 mark**<br>**OR**<br>Incorrect answer but shows 672 divided by 8 = **1 mark**; | 2 | 1. Accept 2 x $10^{25}$ = **2 marks**<br>1. Ignore any numbers after 1.93 |

**Question 10 · A level Paper 2 · June 2022 · Q10.4 · 2 marks**

| | | | |
|---|---|---|---|
| 10.4 | 1. High/increased (concentration of) PSA not always linked to (prostate) cancer<br>**OR**<br>High/increased (concentration of) PSA could be a false positive;<br>2.(Could be) due to urinary infection<br>**OR**<br>(Could be) due to enlarged prostate; | 2<br>(2 x AO2) | 2. Accept 'urine infection'. |


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| | | |
|---|---|---|
| **08.2** | 1. **A**/untreated (type II diabetes) lowest survival (time)<br>**OR**<br>**A**/untreated (type II diabetes) lowers/reduces survival (time compared with control group);<br>2. **B**/metformin increases survival (time) the most<br>**OR**<br>**B**/metformin has the highest survival (time);<br>3. **C**/combined drugs increases survival (time);<br>4. Groups **A and B** have a significant difference (in survival time compared with control);<br>5. Group **C** has no significant difference (in survival time compared with control);<br>6. (In group **C**) other drugs have reduced effect of metformin<br>**OR**<br>**B**/metformin is more effective (treatment/drug) than **C**/combined drugs; | 5 max<br>(5 x AO3) |

1 to 6. Accept descriptions of each group e.g. **A** = no drugs, **B** = metformin, **C** = combined drugs.

4 and 5. Reject 'results are significant' or 'results are not significant' **once,** but **only** where there is no indication that these results are 'different', 'greater', 'reduced' etc.

4 and 5. Accept 'not


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| | |
|---|---|
| 08.4 | **Mark points 1 and 2 are required for maximum marks.**<br>1. Two repeats decreases (risk of) breast cancer;<br>2. Three **and** four repeats increases (risk of) breast cancer;<br>3. No results for more than 4 repeats<br>**OR**<br>No results for 1 repeat<br>**OR**<br>Few repeats (studied);<br>4. Other factors affect (risk of) breast cancer<br>**OR**<br>Named factor (e.g. age) affects (risk of) breast cancer;<br>5. Breast cancers not all same (type);<br>6. Sample size not known; |

| | |
|---|---|
| 4 max (4 x AO3) | Ignore length of investigation and use of a statistical test<br>2. Accept 'more than two repeats'<br>3. Accept only 2,3 and 4 repeats<br>Ignore increase in VNTRs repeats increase risk of breast cancer |


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**Question 13 · A level Paper 2 · November 2020 · Q02.4 · 3 marks**

| | | |
|---|---|---|
| **02.4** | 1. With 16 or fewer than 16 (repeats the association) is significant;<br>2. With 17 or more than 17 (repeats the association) is **not** significant;<br>3. With 16 or fewer than 16 (repeats) there is less than a 5% or less than 0.05 probability of being due to chance<br>**OR**<br>With 17 or more than 17 (repeats) there is more than a 5% or more than 0.05 probability of being due to chance<br>**OR**<br>Explanation of a probability value e.g. 0.30 is a 0.30 or 30% probability of being due to chance;<br>4. With 16 or fewer than 16 (repeats) reject the null hypothesis<br>**OR** | 3 max |
| | With 17 or more (repeats) accept the null hypothesis; | |

If none of the marks is awarded allow principle mark of (prostate) cancer more likely with 16 or less than 16 (repeats) **or** (prostate) cancer less likely with 17 or more than


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| | | |
|---|---|---|
| **05.3** | 1. Only investigated in throat cancer<br>**OR**<br>Might not work for other types of cancer;<br>2. Not all cancers are caused by (increased) methylation (of a tumour suppressor gene)<br>**OR**<br>There are other causes of cancer;<br>3. Only a <u>significant</u> reduction with 20/50/above 10 (µmol)<br>4. Do not know how much EGCG is in green tea;<br>5. Only reduces growth rate (of cancer cells)<br>**OR**<br>No evidence of cancer being cured;<br>6. *In vivo* cells/cells in the body might respond (to EGCG) differently (from those grown *in vitro*); | 3 max |

3. Allow converse, ie no <u>significant</u> effect with 5/10 (µmol)


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