**Cystic Fibrosis, Mutation and Genetic Screening (Topic 2): Edexcel SNAB A Level Biology Exam Questions by Topic**

20 questions · 46 marks · about 55 minutes. Answer in the spaces, then check the mark schemes at the end.

**Question 1 · AS Paper 1 · June 2024 · Q5(a)(ii) · 4 marks**

(ii) Scientists have discovered a property of DNA polymerase that detects and corrects errors in DNA replication.

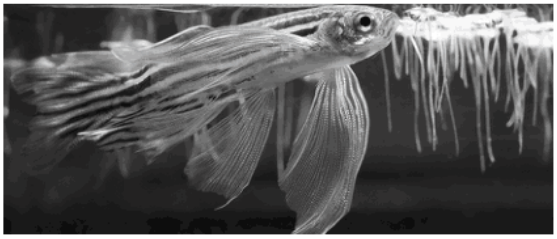
Discuss the possible benefits of this property of DNA polymerase.

**(4)**


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(b) Zebrafish with different phenotypes have been produced by introducing gene mutations into the zebrafish genome.

Two of these phenotypes, spots and long fin, are described in the table.

| Phenotype name | Photograph | Description |
|---|---|---|
| spots |  (Source: © Grigorev Mikhail/Shutterstock) | melanin produced in spots instead of stripes |
| long fin |  (Source: © Grigorev Mikhail/Shutterstock) | long fins instead of short fins |

In an investigation, the inheritance of these two phenotypes was studied.

In cross 1, one parent was homozygous for stripes and the other parent was homozygous for spots.

In cross 2, one parent was homozygous for short fins and the other parent was homozygous for long fins.

The table shows the results of this investigation.

| Cross | Genotype of parent 1 | Genotype of parent 2 | Phenotype of the offspring |
|---|---|---|---|
| 1 | homozygous for stripes and short fins | homozygous for spots and short fins | all had stripes and short fins |
| 2 | homozygous for stripes and short fins | homozygous for stripes and long fins | all had stripes and long fins |

(i) State what is meant by the term gene mutation.

**(1)**

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**Question 3 · A level Paper 2 · June 2019 · Q3(b)(ii) · 2 marks**

(ii) Explain why different mutations in the CFTR gene can lead to differences in the severity of the symptoms of cystic fibrosis.

**(2)**

**Question 4 · A level Paper 1 · June 2017 · Q2(c)(i) · 3 marks**

(c) Some epithelial cells in the lungs secrete mucus. If the mucus is too 'sticky', it cannot be easily removed from the lungs.

Other epithelial cells in the lungs contain CFTR proteins in their cell surface membranes.

(i) Describe the role of the CFTR protein in ensuring that the mucus produced in the lungs has the right consistency.

**(3)**


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**Question 5 · AS Paper 1 · June 2018 · Q8(b) · 2 marks**

(b) More than 1500 mutations have been found for the gene that codes for the production of the CFTR channel protein.

Some of these mutations cause cystic fibrosis by affecting the production or functioning of the CFTR channel protein.

If the functioning of the CFTR channel protein is impaired, thicker mucus is produced in the lungs.

Explain why thicker mucus is produced if the functioning of the CFTR channel protein is impaired.

**(2)**

**Question 6 · A level Paper 2 · June 2017 · Q3(a) · 3 marks**

**3** Cystic fibrosis is a condition that affects breathing.

(a) Explain why cystic fibrosis affects the rate of oxygen uptake in the lungs.

**(3)**


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**Question 7 · AS Paper 1 · June 2022 · Q5(a) · 1 mark**

**5** Cystic fibrosis can result from mutations in the CFTR gene.

These mutations affect the function of the CFTR protein channel in the epithelial cells of several organ systems, including the respiratory system.

(a) Name another organ system that would be affected by a CFTR gene mutation.

**(1)**

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**Question 8 · A level Paper 2 · June 2023 · Q4(c) · 3 marks**

(c) Genetic screening can be used to test for conditions such as cystic fibrosis.

Explain why cystic fibrosis affects digestion.

**(3)**

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**Question 9 · AS Paper 1 · October 2020 · Q8(d)(ii) · 5 marks**

(ii) Doctors give dietary supplements and digestive enzymes to children with cystic fibrosis.

Dietary supplements include carbohydrates, proteins and lipids, as well as vitamin and mineral supplements.

Explain why these children would be given dietary supplements and digestive enzymes.

**(5)**


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**Question 10 · A level Paper 2 · June 2023 · Q4(a) · 4 marks**

**4** There are different types of genetic screening available.

Each year, about 30 000 prenatal genetic screening tests are carried out.

(a) Compare and contrast the use of pre-implantation genetic diagnosis (PGD) and amniocentesis.

**(4)**

**Question 11 · AS Paper 1 · June 2018 · Q4(b)(iii) · 1 mark**

(iii) An embryo, created by IVF, can be screened before it is placed in the mother's uterus.

Name this type of genetic screening.

**(1)**

**Question 12 · AS Paper 1 · June 2023 · Q4(b)(ii) · 1 mark**

(ii) An embryo, created by IVF, can be screened for Tay-Sachs disease before being placed in the mother's uterus.

Name this type of genetic screening.

**(1)**


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**Question 13 · AS Paper 1 · June 2023 · Q4(b)(iii) · 2 marks**

(iii) Explain why the mother may prefer this type of prenatal screening to amniocentesis.

**(2)**


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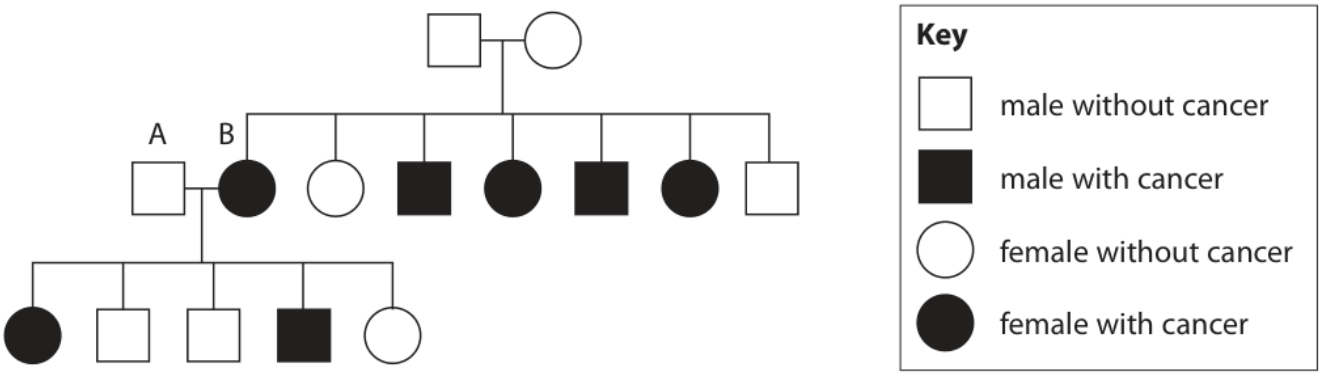
**Question 14 · AS Paper 1 · June 2024 · Q5(b) · 2 marks**

(b) Some people have DNA polymerase that cannot detect and correct errors in DNA replication.

These people have an increased risk of developing cancer.

The increased risk of cancer occurs when a person inherits two faulty recessive alleles of the DNA polymerase gene.

The pedigree diagram shows the development of cancer in one family.




Individuals A and B wish to have another child.

Explain why they may choose pre-implantation genetic diagnosis (PGD).

**(2)**


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**Question 15 · A level Paper 2 · June 2024 · Q3(c) · 1 mark**

(c) Radiation has leaked from accidents at other nuclear power stations. This has led to mutations in the developing embryos of pregnant women.

Prenatal genetic screening can be offered to check for certain mutations.

Give a reason why some pregnant women may choose chorionic villus sampling rather than amniocentesis.

**(1)**

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**Question 16 · AS Paper 1 · June 2016 · Q1(b) · 1 mark**

(b) Tay-Sachs disease can be detected during pregnancy.

Name the prenatal test that could be used to detect Tay-Sachs disease at 11 weeks of pregnancy.

**(1)**

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**Question 17 · AS Paper 1 · June 2016 · Q1(c) · 3 marks**

(c) Explain why this couple may choose not to have this test.

**(3)**

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**Question 18 · AS Paper 1 · June 2018 · Q4(b)(iv) · 2 marks**

(iv) Explain **one** ethical issue relating to the use of prenatal genetic screening.

**(2)**

**Question 19 · AS Paper 1 · June 2022 · Q5(c)(iv) · 3 marks**

(iv) Discuss the ethical issues relating to the use of genetic screening during pregnancy.

**(3)**


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**Question 20 · A level Paper 2 · June 2023 · Q4(b) · 2 marks**

(b) Describe **two** different social issues related to the use of PGD.

**(2)**


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## Mark schemes

**Question 1 · AS Paper 1 · June 2024 · Q5(a)(ii) · 4 marks**

| Question Number | Answer |
|---|---|
| 5(a)(ii) | An answer that makes reference to the following:<br>• (this property of DNA polymerase) would reduce the number of {mutations / faulty alleles} (1)<br>• (ensuring) the correct {base sequence / mRNA sequence / allele} is produced (1)<br>• (ensuring) the correct {amino acid sequence / polypeptide / protein} is produced (1)<br>• reducing the chance of {cancer / genetic disease} (1) |

| Additional Guidance | Mark |
|---|---|
| ALLOW errors in DNA replication cause {mutations / change in base sequence / changes in bases}<br><br>ALLOW faulty bases are replaced<br>ALLOW the (original) base sequence is {not changed / restored}<br>ignore errors are corrected<br><br>ALLOW incorrect base sequence can result in the production of a {polypeptide with a different primary structure / non-functioning protein}<br>ALLOW description of change in active site shape changing {number of enzyme-substrate complexes formed / rate of reaction}<br><br>ALLOW uncontrolled cell division<br>ignore disease unqualified | **(4)** |


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**Question 2 · A level Paper 3 · June 2023 · Q6(b)(i) · 1 mark**

| Question number | Answer |
|---|---|
| 6(b)(i) | • **change** in the **base sequence** in the {**DNA / gene**} |

| Additional guidance | Mark |
|---|---|
| | (1) |

**Question 3 · A level Paper 2 · June 2019 · Q3(b)(ii) · 2 marks**

| Question Number | Answer |
|---|---|
| **3(b)(ii)** | An explanation that makes reference to two of the following:<br>• (different mutations) will have different effects on the protein produced (1)<br>• chloride ion transport affected by the extent of changes to the (CFTR) protein (1)<br>• varying the { stickiness / thickness } of the mucus (1) |

| Additional Guidance | Mark |
|---|---|
| ALLOW absence of protein / different { folding / tertiary structure / shape } of the protein<br>ALLOW faulty CFTR protein has less impact on chloride ion transport than a missing CFTR protein | **(2)** |


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**Question 4 · A level Paper 1 · June 2017 · Q2(c)(i) · 3 marks**

| Question Number | Answer |
|---|---|
| 2(c)(i) | A description that makes reference to three of the following:<br>• chloride ions leave cells (through the CFTR channel protein) (1)<br>• sodium ions leave the cells (following the chloride ions) (1)<br>• increasing the solute concentration in the mucus (1)<br>• water moves out of the cells by osmosis (into the mucus) (1) |

| Additional Guidance | Mark |
|---|---|
| NOT active transport of chloride ions<br>ALLOW chloride ions move into the mucus<br>ALLOW NaCl, $Na^+$ or $Cl^-$ instead of solute<br>ALLOW description of osmosis | 3 |

**Question 5 · AS Paper 1 · June 2018 · Q8(b) · 2 marks**

| Question Number | Answer |
|---|---|
| 8(b) | An explanation that makes reference to two of the following:<br>• chloride ions cannot {leave the cell / enter mucus} (through CFTR protein channel) (1)<br>• sodium ions do not move { out of the cells / into the mucus } (1)<br>• therefore water {moves into cells / moves out of mucus} by osmosis (1) |

| Additional Guidance | Mark |
|---|---|
| ALLOW water does not move into mucus by osmosis | (2) |


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**Question 6 · A level Paper 2 · June 2017 · Q3(a) · 3 marks**

| Question number | Answer |
|---|---|
| **3(a)** | An explanation that makes reference to three of the following:<br>• { thick / sticky / viscous } mucus (1)<br>• (accumulation of mucus) which cannot be moved by cilia (1)<br>• restricting air flow through { bronchioles / bronchi } (1)<br>• {increases diffusion distance / reduces surface area for gas exchange } in the alveoli (1) |

| Additional guidance | Mark |
|---|---|
| IGNORE 'airways'<br>ALLOW narrowing of bronchioles | (3) |

**Question 7 · AS Paper 1 · June 2022 · Q5(a) · 1 mark**

| Question Number | Answer | Additional Guidance | Mark |
|---|---|---|---|
| **5 (a)** | digestive (system) / reproductive (system) | | **(1)** |


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**Question 8 · A level Paper 2 · June 2023 · Q4(c) · 3 marks**

| Question Number | Answer |
|---|---|
| 4(c) | An explanation that makes reference to three of the following:<br>• (produces) sticky mucus (1)<br>• (which) { blocks / reduces } flow of {pancreatic juice / enzyme from pancreas } (1)<br>• (so) reducing digestion (1)<br>• {blocks / reduces } flow of bile (1) |

| Additional Guidance | Mark |
|---|---|
| ALLOW: thick/thicker/viscous mucus<br>ALLOW: blocks / reduces lumen diameter of pancreatic duct<br>ALLOW a named enzyme<br>ALLOW: food not digested, food not broken down<br>ALLOW: blocks / reduces lumen diameter of bile duct | [3] |


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**Question 9 · AS Paper 1 · October 2020 · Q8(d)(ii) · 5 marks**

| Question Number | Answer |
|---|---|
| 8(d)(ii) | An explanation which makes reference to five of the following:<br>• cystic fibrosis causes the production of {thicker / stickier} mucus (1)<br>• which blocks the pancreatic duct / prevent pancreas enzymes reaching intestine (1)<br>• {reduces / prevents} enzymes digesting {carbohydrates / lipids / proteins} (in intestines) (1)<br>• (resulting in) reduced {absorption / concentration} of products of digestion into the {blood / lymph} (1)<br>• linkage of reduced {amino acids / vitamins / minerals} to slower growth rate (1)<br>• {dietary supplements / digestive enzymes} would increase growth rate (1) |

| Additional guidance | Mark |
|---|---|
| ALLOW converse for given digestive enzymes for mp3+4<br>ALLOW reduced digestion of food by enzymes<br>e.g. amino acids / glucose / fatty acids / glycerol / vitamins / minerals | (5) |


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**Question 10 · A level Paper 2 · June 2023 · Q4(a) · 4 marks**

| Question Number | Answer |
|---|---|
| 4(a) | An answer that makes reference to the following:<br><br>Similarity<br><br>• (both) look for any genetic condition (so parents can make informed decisions) (1)<br><br>Differences<br><br>• PGD before implantation / amniocentesis after implantation (1)<br><br>• (increased) risk of miscarriage with amniocentesis (1)<br><br>• embryo tested in PGD / amniotic fluid tested in amniocentesis (1) |

| Additional Guidance | Mark |
|---|---|
| ALLOW testing occurs earlier in PGD / amniocentesis occurs during pregnancy<br><br>ALLOW {reduced /no} risk of miscarriage with PGD<br>ALLOW selecting embryo for implantation in PGD<br><br>ALLOW cells from the 'amniotic fluid' | [4] |

**Question 11 · AS Paper 1 · June 2018 · Q4(b)(iii) · 1 mark**

| Question Number | Answer | Additional Guidance | Mark |
|---|---|---|---|
| 4 (b)(iii) | • Pre-implantation genetic diagnosis / PGD / PIGD | ALLOW Pre-implantation genetic screening / PGS | (1) |


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**Question 12 · AS Paper 1 · June 2023 · Q4(b)(ii) · 1 mark**

| Question Number | Answer | Additional Guidance | Mark |
|---|---|---|---|
| 4 (b)(ii) | • pre-implantation genetic diagnosis / PGD / PIGD | ALLOW pre-implantation genetic screening / PGS / PGT | (1) |

**Question 13 · AS Paper 1 · June 2023 · Q4(b)(iii) · 2 marks**

| Question Number | Answer | Additional Guidance | Mark |
|---|---|---|---|
| 4(b)(iii) | An answer that makes reference to the following:<br>• fewer ethical issues (with PGD) as the embryo has not been implanted / amniocentesis carried out at a later stage (1)<br>• no risk of miscarriage from PGD / risk of miscarriage from amniocentesis (1) | e.g. embryo in earlier stage of development / PGD carried out at an earlier stage | (2) |

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**Question 14 · AS Paper 1 · June 2024 · Q5(b) · 2 marks**

| Question Number | Answer |
|---|---|
| 5(b) | An explanation that makes reference to two of the following:<br>• {parent A / father} is {a carrier / heterozygous} / {parent B / mother} has cancer (1)<br>• PGD allows parent to select embryo that {is not homozygous for this (faulty) allele / does not have the faulty allele / produces the functioning DNA polymerase} (1)<br>• (PGD has) no risk of miscarriage (1) |

| Additional Guidance | Mark |
|---|---|
| ALLOW because some of their children have cancer<br>ignore gene<br>ALLOW to know if their child {has a faulty allele / is a carrier / has recessive genotype}<br>ALLOW to reduce risk of child developing cancer<br>ignore to choose a child without {condition / cancer / genetic disorders unqualified}<br>ignore PGD is non-invasive<br>ignore abortion | **(2)** |


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**Question 15 · A level Paper 2 · June 2024 · Q3(c) · 1 mark**

| Question Number | Answer |
|---|---|
| 3(c) | An answer that makes reference to one of the following:<br>• earlier in pregnancy when test can be carried out (1)<br>• results available sooner (due to being carried out earlier in pregnancy) (1) |

| Additional Guidance | Mark |
|---|---|
| ALLOW reference to number of weeks to show CVS is done earlier in pregnancy (10-14 weeks) than amniocentesis (15-20 weeks)<br>ALLOW longer time to consider the outcome<br>IGNORE reference to miscarriage risk | 1 |

**Question 16 · AS Paper 1 · June 2016 · Q1(b) · 1 mark**

| Question Number | Answer | Additional Guidance | Mark |
|---|---|---|---|
| 1(b) | chorionic villus sampling | Accept CVS<br>Accept phonetic spelling<br>Do not accept chronic villus sampling | (1) |


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**Question 17 · AS Paper 1 · June 2016 · Q1(c) · 3 marks**

| Question Number | Answer |
|---|---|
| 1(c) | An explanation that makes reference to three of the following:<br>• test result may be inaccurate (1)<br>• (increased) risk of miscarriage (due to the procedure) (1)<br>• false positive may lead to termination of healthy fetus (1)<br>• { prenatal testing / abortion } against values or beliefs of the parents (1) |

| Additional Guidance | Mark |
|---|---|
| Ignore reference to amniocentesis<br>Allow false positives / false negatives<br>Allow spontaneous abortion<br>Allow 'parents do not want to know' | (3) |


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**Question 18 · AS Paper 1 · June 2018 · Q4(b)(iv) · 2 marks**

| Question Number | Answer |
|---|---|
| 4 (b)(iv) | An explanation that makes reference to two of the following:<br><br>either<br>• it may result in a choice of an abortion (1)<br>• it is unethical to cause the death of a foetus (1)<br><br>or<br>• (risk of) {incorrect result / false positive / false negative } (1)<br>• healthy foetus could be aborted / parents not prepared for child with {genetic disease / achondroplasia} (1) |

| Additional Guidance | Mark |
|---|---|
| ALLOW can result in embryos being discarded<br><br>ALLOW unethical to destroy a potential human being<br><br>ALLOW can result in healthy embryos being discarded | (2) |

**Question 19 · AS Paper 1 · June 2022 · Q5(c)(iv) · 3 marks**

| Question Number | Answer | Additional Guidance | Mark |
|---|---|---|---|
| 5(c)(iv) | An answer that makes reference to the following:<br>• (1-2%) risk of a miscarriage (1)<br>• risk of false positive result (1)<br>• cystic fibrosis patients can have high {quality of life / life expectancy} due to treatments available (1) | ALLOW risk of harm to the fetus<br>e.g. resulting in termination of pregnancy when fetus was healthy<br>ALLOW some believe that aborting a fetus due to a genetic condition is {unethical / wrong as the foetus has the potential to become a human} | (3) |

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**Question 20 · A level Paper 2 · June 2023 · Q4(b) · 2 marks**

| Question Number | Answer |
|---|---|
| **4(b)** | A description that makes reference to two of the following:<br>• selecting one embryo over another / loss of a potential life (1)<br>• risk of identifying other conditions / selecting for other characteristics (1)<br>• {costly / emotional strain } as not always accurate (1) |

| Additional Guidance | Mark |
|---|---|
| ALLOW issues concerning discarding of spare embryos<br>ALLOW 'designer baby' argument<br>ALLOW 'false positives' for 'not always accurate' | [2] |


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