

# Inheritance, cystic fibrosis and genetic screening: practice questions

7 questions · 26 marks · mark scheme at the back

 For Pearson Edexcel International AS Biology XBI11 and International A Level Biology YBI11. **A Level only** is IA2 content (Units 4 to 6), not needed for the International AS.

Name .....

Total ..... / 26

**1** UNIT 1 STYLE [1 mark]

Two people who both have blood group AB (genotype  $I^A I^B$ ) have a child. Which blood group could the child NOT have?

- A. ☐ Group A
- B. ☐ Group B
- C. ☐ Group AB
- D. ☐ Group O

**2** UNIT 1 STYLE [4 marks]

The table describes the members of one family in which some individuals have a genetic condition. Use the table to deduce whether the allele that causes the condition is dominant or recessive. Explain your answer.

| Family with a genetic condition |        |              |             |
|---------------------------------|--------|--------------|-------------|
| Individual                      | Sex    | Condition    | Parents     |
| I-1                             | Male   | Not affected | —           |
| I-2                             | Female | Not affected | —           |
| II-1                            | Female | Affected     | I-1 and I-2 |
| II-2                            | Male   | Not affected | I-1 and I-2 |

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3

UNIT 1 STYLE

[3 marks]

Red-green colour blindness is caused by a recessive allele ( $X^b$ ) on the X chromosome. A man with normal colour vision and a woman who is a carrier of the allele plan to have a child. Draw a genetic diagram and give the probability that their first child is a colour-blind boy.

Answer = .....

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4

UNIT 1 STYLE

[5 marks]

Explain how a mutation in the gene for the CFTR protein leads to the symptoms of cystic fibrosis in the gaseous exchange system and the digestive system.

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5

UNIT 1 STYLE

[4 marks]

Compare and contrast amniocentesis and chorionic villus sampling.

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**6** UNIT 1 STYLE

**[3 marks]**

A couple who are both carriers of the cystic fibrosis allele decide to have a child by IVF with pre-implantation genetic diagnosis (PGD). Describe how PGD is carried out.

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**7** UNIT 1 STYLE

**[6 marks]**

Discuss the ethical and social issues relating to prenatal genetic screening, from religious, moral and social viewpoints.

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## Mark scheme

One mark for each point unless a maximum is given. Words in brackets are not needed for the mark.

### 1. [1]

**D** D. The possible child genotypes are  $I^A I^A$  (A),  $I^A I^B$  (AB) and  $I^B I^B$  (B). Neither parent has an  $I^O$  allele, so group O is not possible.

### 2. [4]

- the allele is recessive (1)
- I-1 and I-2 are not affected but have an affected child, II-1 (1)
- if the allele were dominant, one of the parents would have to be affected (1)
- so I-1 and I-2 must both be heterozygous / carriers (1)
- II-1 is homozygous recessive (1)
- Max 4

### 3. [3]

- parents  $X^B Y$  and  $X^B X^b$ ; gametes  $X^B$ , Y and  $X^B$ ,  $X^b$  (1)
- offspring  $X^B X^B$ ,  $X^B X^b$ ,  $X^B Y$ ,  $X^b Y$  (1)
- probability 1 in 4 / 25 % / 0.25 (1)

### 4. [5]

- mutation changes the sequence of amino acids / protein structure so the chloride channel protein does not work (1)
- chloride ions not secreted out of the cells, so less water leaves the cells by osmosis (1)
- mucus is thick and sticky (1)
- gaseous exchange: airways blocked / mucus traps bacteria and causes infections, reducing the surface area for gas exchange (1)
- digestive: pancreatic duct blocked so enzymes cannot reach the small intestine, so less digestion and absorption (1)
- mucus cannot be moved by cilia / bacteria are trapped and not removed (1)
- Max 5

### 5. [4]

- similarity: both are prenatal tests / involve inserting a needle guided by ultrasound / carry a risk of miscarriage (1)
- similarity: both obtain fetal cells for DNA or chromosome analysis (1)
- difference: amniocentesis takes amniotic fluid, CVS takes placental cells / chorionic villi (1)
- difference: amniocentesis is done later in pregnancy (about 15 weeks), CVS earlier (about 11 weeks) (1)
- difference: CVS gives results earlier (1)
- Max 4

### 6. [3]

- embryos are made by in vitro fertilisation and allowed to develop to about 8 cells (1)
- one or two cells are removed from each embryo and the DNA is tested for the cystic fibrosis allele (1)
- only embryos without two copies of the allele are implanted into the uterus (1)

## 7. [6]

- religious: some religious groups believe life is sacred from conception, so termination after screening is wrong (1)
- religious: others accept screening to avoid severe suffering / views differ within religions (1)
- moral: parents' right to information and choice / autonomy (1)
- moral: every life has value / a fetus has a right to life / disability rights (1)
- social: pressure on parents to screen or to terminate / stigma towards people with the disorder (1)
- social: discrimination, for example by insurers or employers / privacy of genetic information (1)
- social: cost and access / reduces burden on families and health services (1)
- conclusion that weighs both sides (1)
- Max 6