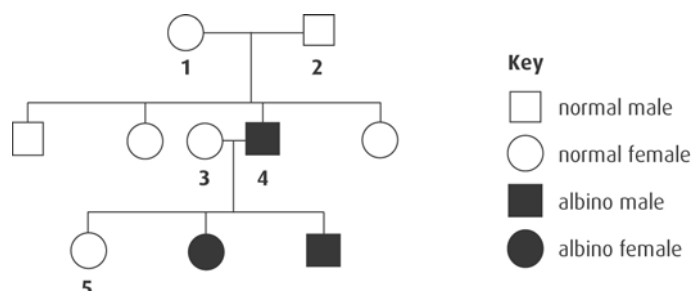


## Exemplar exam questions – Chapter 4, *Genetics I*

### Structured questions

#### Exemplar question 1

Albinism is an autosomal, recessive condition in which the skin pigment melanin does not form. The pedigree chart shown below is of a family that has several members with albinism.



- a Give the genotypes of the parents **1** and **2**, the female **3** and the male **4**, who has albinism. (4)
- b What is the probability that female **5** is a carrier of albinism? (1)
- c How many members of the family are definitely heterozygous for this allele? (1)

#### Student answer

- a Parents **1** and **2** must be heterozygous because they have produced children both with and without albinism.

If A is the normal allele and a is the recessive allele, both **1** and **2** must be Aa.

Female **3** does not have albinism but her husband does. He must be aa but as they have children with and without albinism, she must be Aa because her second daughter and son have albinism.

- b Female **5** must be a carrier because she has one a allele from her father and one A allele from her mother. She must be Aa.
- c Individuals **1** and **2** are definitely heterozygous and individuals **3** and **5** are definitely heterozygous.

### Commentary

This student has a good understanding of the questions and has given correct answers but he or she has not always responded clearly to what has been asked in the question.

- a** The answer is correct but the student has not clearly stated what the genotype of person **4** (who is only referred to as a 'husband') actually is, and so would lose a mark. This answer would be awarded 3 marks.
- b** A good answer that would be awarded 1 mark.
- c** What the student has written is correct but the number of individuals who are definitely heterozygous is not stated (4). This answer would be awarded 0 marks.

**Total marks awarded:** 4 out of 6

### Exemplar question 2

Nephrogenic diabetes insipidus is a rare genetic disorder, which has been shown to be caused by a mutation in the gene coding for a hormone receptor protein. The mutant form of the allele has a 'stop' codon instead of the codon for the amino acid tryptophan in the middle of the gene.

- a** State the type of mutation that converts one codon into another. (1)
- b** Predict the effect of converting this codon in the middle of the gene into a 'stop' codon. (2)

### Student response

- a** a point mutation
- b** A stop codon will cause mRNA to detach from the ribosome before the whole polypeptide has been made. This will produce a shortened protein, which will not be effective as a receptor.

### Commentary

This is a good answer to a structured-style question. The facts are given clearly with no excess information that is not required.

**Total marks awarded:** 3 out of 3

### Exemplar question 3

By reference to the ABO blood group system, explain what is meant by:

- a** multiple alleles (3)
- b** codominance (2)
- c** a recessive allele. (2)

**Student response**

- a** Each person has two copies of alleles for each of their genes. Some genes have two different alleles, e.g. B and b for brown and blue eyes, but others have more than two. In the ABO blood group system there are three possible alleles A, B and i. Each person has two of these.
- b** A dominant allele is one that is expressed in a person's phenotype even if there is only one copy of it. A recessive allele is not apparent in a phenotype unless there are two copies. Codominance occurs when two dominant alleles are present and both affect a phenotype. In ABO blood groups A and B are both dominant and if a person has both they are blood group AB.
- c** A recessive allele only affects a phenotype if there are two copies of it. In blood groups, a person must have two i alleles to be blood group O.

**Commentary**

This student clearly understands the genetics of ABO blood groups but could smarten up his or her answer by taking care with terminology.

In IB exams, the standard notation  $I^B$ ,  $I^A$  and i should always be used for blood group genotypes. Section **a** has been answered clearly and concisely. **b** is worth 2 marks and could be answered more concisely. If the student had read the whole question before responding, he or she would have seen that the definition of a recessive allele (correctly given) should be included in **c** and not **b**.

**Total marks awarded:** 6 out of 7