

- 1 Guinea pigs, *Cavia porcellus*, vary in the length and colour of their fur.

Fig. 1.1 shows a guinea pig with short black fur.



Fig. 1.1

Two genes that determine the length and colour of the fur occur at the **A/a** locus and the **B/b** locus. These two gene loci are on separate autosomal chromosomes.

- The allele **A** results in short fur.
- The allele **a** results in long fur.
- **A** is dominant to **a**.
- The allele **B** results in black fur.
- The allele **b** results in chocolate fur.
- **B** is dominant to **b**.

- (a) (i) List **all** the possible genotypes of a guinea pig with short black fur.

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- (ii) A test cross could be used to determine the genotype of a female guinea pig with short black fur.

Describe the **phenotype** of the male guinea pig that could be used to carry out this test cross.

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(b) A black guinea pig with long fur that was homozygous at both loci was crossed with a chocolate guinea pig with short fur that was homozygous at both loci. The F1 offspring of this cross had short black fur. F1 offspring were mated together to produce the F2 offspring.

- Complete the Punnett square to:
- show the cross between the F1 offspring
 - predict the F2 offspring genotypes.
- You should include the gametes in your answer.

State the ratio of F2 offspring phenotypes. You should include a key to link phenotypes to genotypes.

ratio of F2 offspring phenotypes:

[4]

(c) Some genes in guinea pigs are structural genes and some are regulatory genes.

Describe the difference between a structural gene and a regulatory gene.

[2]

4 The wolf, *Canis lupus*, lives in North America. Wolves may have a grey or a black coat colour. The colour of an individual wolf depends on the DNA it inherits at the *CPD103* gene locus.

- Wolves inherit two copies of *CPD103*, one from each parent.
- Wolves that inherit one copy of the black form of the *CPD103* gene have a black coat.

(a) State the term used to describe:

- an organism that has two copies of each gene
- a form of a gene
- a form of a gene that gives a phenotypic effect in a heterozygote.

[3]

In addition to producing black coat colour, the protein coded for by the *CPD103* gene also defends against infectious lung disease.

Canine distemper virus (CDV) causes serious lung disease in wolves. Wolves that have been previously infected by CDV have antibodies against CDV (anti-CDV antibodies) in their blood.

(b) CDV can be passed from domestic dogs to wolves.

- Domestic dogs are more numerous in the southern part of the area occupied by wolves.
- Domestic dogs are less numerous in the northern part of the area occupied by wolves.
- The relative frequency of black wolves compared to grey wolves increases from the north to the south of the area they occupy.

Explain how natural selection causes this trend in the distribution of black wolves.

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(c) Several different populations of wolves were compared.

Fig. 4.1 shows the relationship between the percentage of wolves in a population that have anti-CDV antibodies in their blood and the percentage of wolves in that population that are black. The line of best fit was calculated after comparing the different populations of wolves.

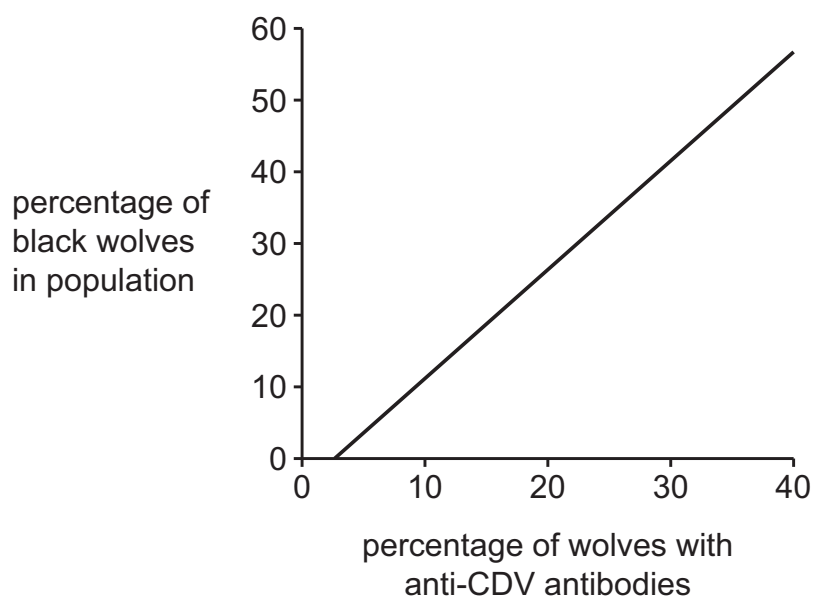


Fig. 4.1

With reference to Fig. 4.1, state the relationship between the percentage of wolves with anti-CDV antibodies and wolf coat colour, **and** suggest reasons for this relationship.

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[Total: 11]

- 3 Porphyria is a group of rare genetic diseases in which molecules called porphyrins accumulate in the body.

Three examples of porphyria are:

- X-linked protoporphyria
- variegate porphyria
- congenital erythropoietic porphyria (CEP).

- (a) X-linked protoporphyria is caused by a mutant allele located on the X chromosome.

Fig. 3.1 shows the pattern of inheritance of X-linked protoporphyria in one family.

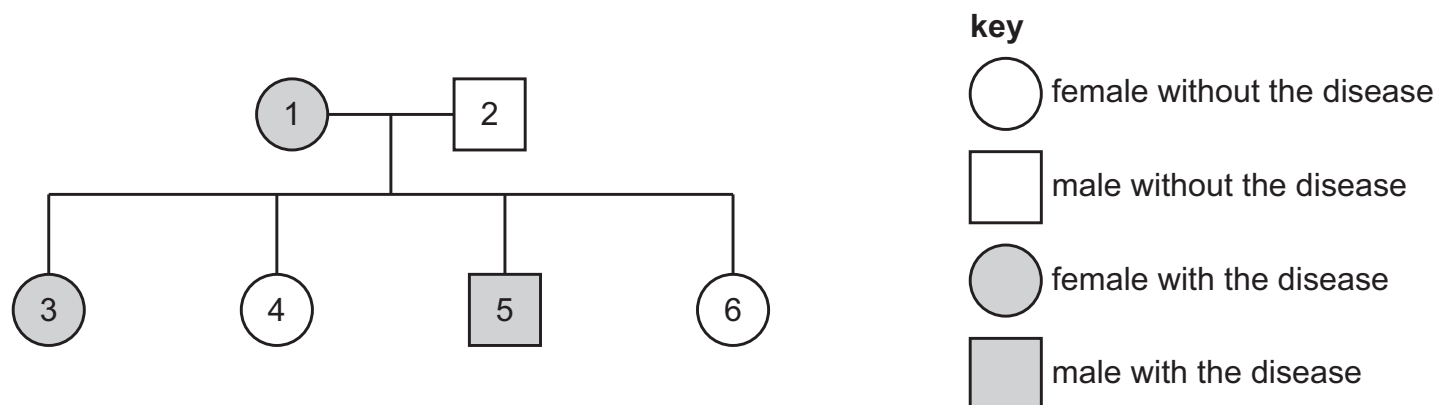


Fig. 3.1

X-linked protoporphyria is described as a sex-linked disease.

Use the information in Fig. 3.1 to make **one** other conclusion about the pattern of inheritance of X-linked protoporphyria.

Give evidence to support your conclusion.

conclusion

evidence

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[2]

(b) Variegate porphyria occurs in 1 in 100 000 people in Europe.

In the 1600s, approximately 100 Dutch people migrated from Europe to South Africa.

The population of people of Dutch descent in South Africa is now 2.5 million. Variegate porphyria occurs in 1 in 1000 people in this population.

Suggest **and** explain why variegate porphyria is more common among people of Dutch descent in South Africa than among people in Europe.

[3]

(c) Microarrays can be used to detect various forms of porphyria caused by mutant alleles.

Describe how microarrays can be used to detect a disease by analysing gene expression.

[4]

- (d) The nucleotide sequences of alleles that cause rare genetic diseases, such as the various forms of porphyria, are stored in a database.

State **one** benefit of having a database of nucleotide sequences for alleles that cause rare diseases.

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- (e) Congenital erythropoietic porphyria (CEP) is caused by a substitution mutation in a gene coding for an enzyme.

The mutant CEP allele codes for an enzyme with little or no function.

Scientists used gene editing in the laboratory to correct the nucleotide sequence of the mutant allele in cultured human cells.

The scientists introduced three molecules to the cells:

- an enzyme called Cas9 that causes breaks in DNA strands
- guide RNA (gRNA), attached to Cas9, that is complementary to the mutant CEP allele
- a short length of DNA, known as template DNA, that can replace the section of the allele where the substitution mutation has occurred.

The repair mechanism within the cell allows the template DNA to be inserted so that the mutation is corrected.

Fig. 3.2 shows part of the nucleotide sequence in the mutated CEP allele before and after the gene editing procedure.

T A T G G C T C G $\xrightarrow{\text{gene editing}}$ T A C G G C T C G

Fig. 3.2

In the future, this gene editing procedure may be used in a person with CEP to prevent the accumulation of porphyrins in the body.

Suggest **and** explain how this gene editing procedure will prevent the accumulation of porphyrins in a person with CEP without damaging other genes.

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