

1 Various internal and external factors can affect the phenotype of humans.

(a) Explain what is meant by the term **phenotype**.

(2)

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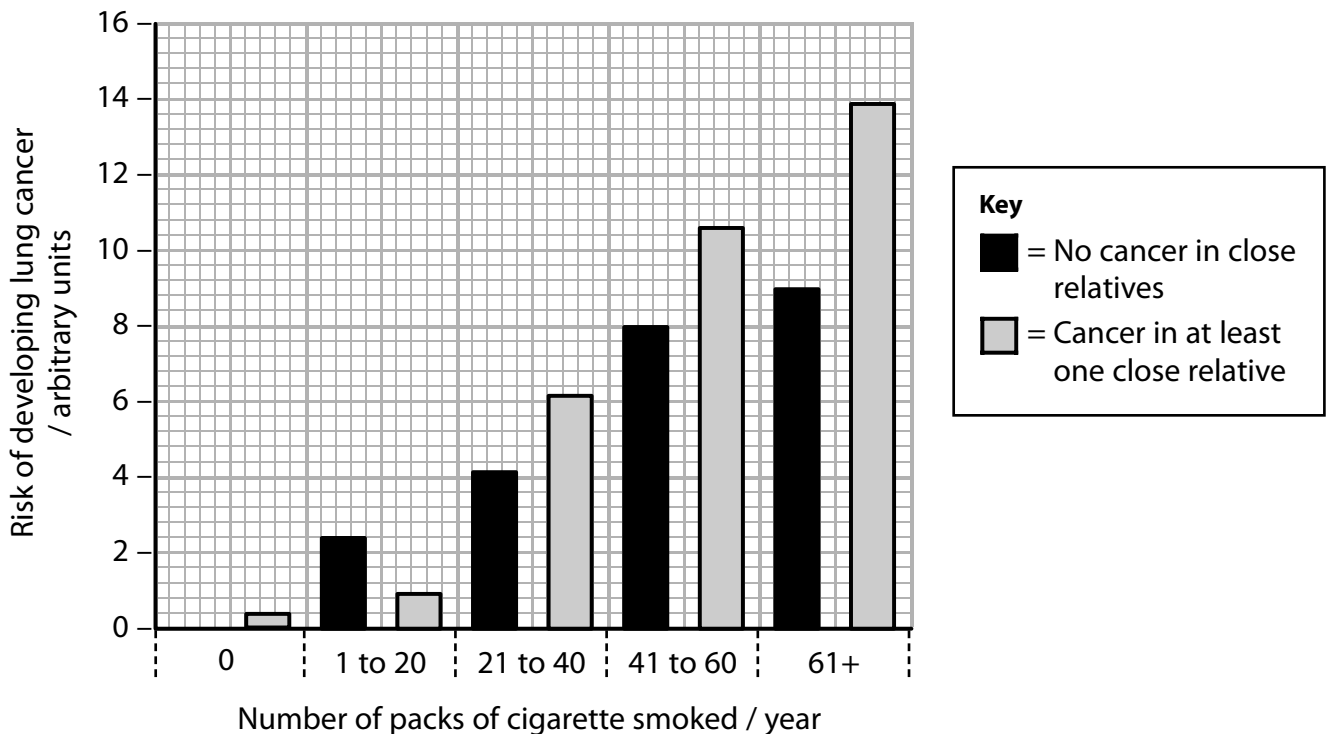
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(b) A survey was carried out to investigate the factors affecting the risk of developing lung cancer. In this survey, people were asked how many packs of cigarettes they smoked per year. They were also asked whether a close relative (a brother, sister or parent) had developed some form of cancer.

The results of the survey are shown in the graph below.

A risk of developing lung cancer of 0 arbitrary units means that there is no increased risk.



(i) Identify the control group in this survey and suggest why they were included. (2)

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(ii) State the phenotype being investigated. (1)

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(iii) Describe the effect of smoking on the increased risk of developing lung cancer, for the people who had close relatives who developed cancer. (2)

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(iv) Use evidence from the graph to support the suggestion that genetic factors may influence the risk of developing lung cancer. (2)

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(v) Give **one** piece of evidence from the graph which suggests that genetic factors may **not** influence the risk of developing lung cancer.

(1)

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(vi) Give **one** piece of evidence from the graph which suggests that external factors may influence the risk of developing lung cancer.

(1)

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(Total for Question 1 = 11 marks)

2 The phenotype of an individual is dependent on a variety of factors.

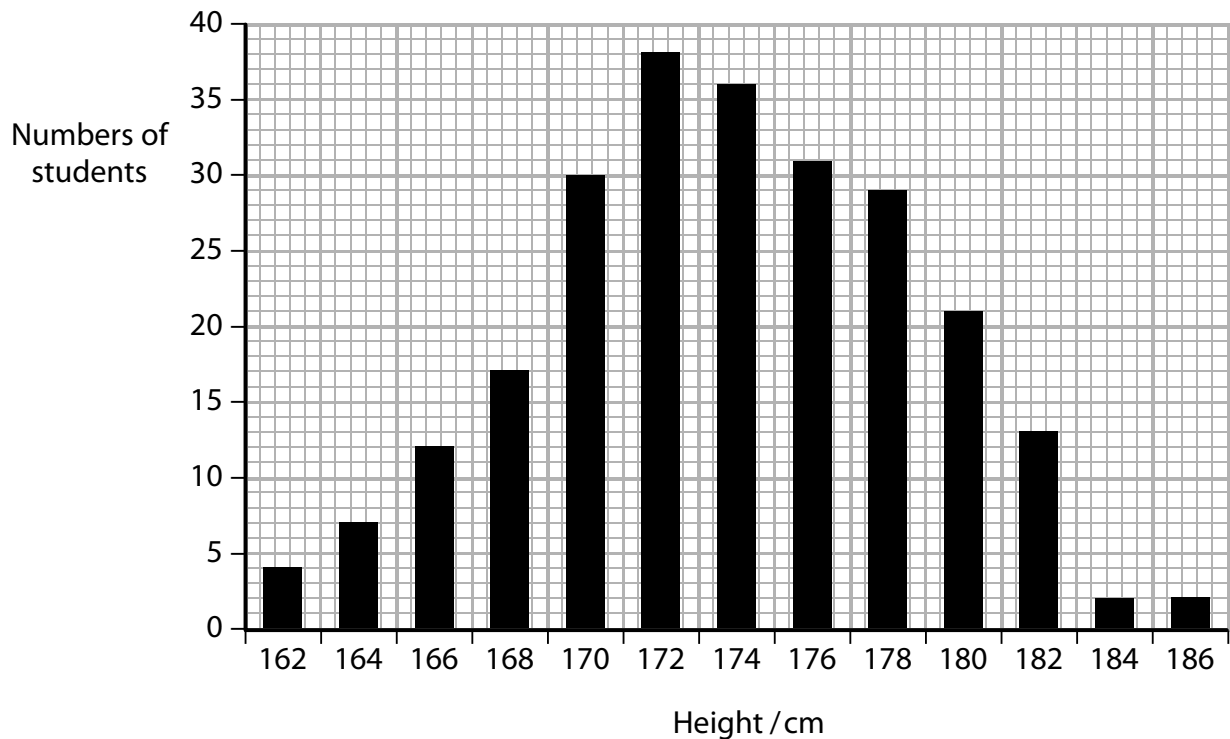
(a) (i) Complete the following sentence by writing the most appropriate word or words on the dotted lines.

(2)

In polygenic inheritance, the phenotypes are affected by forms of genes called found at many on chromosomes.

(ii) The graph below shows some data on the numbers of students of different heights.

Height is an example of polygenic inheritance.



State the heights that represents the median and the mode for this group of students.

(2)

Median height cm

Mode height cm

- (b) (i) Complete the following short passage by writing the most appropriate word or words on the dotted lines.

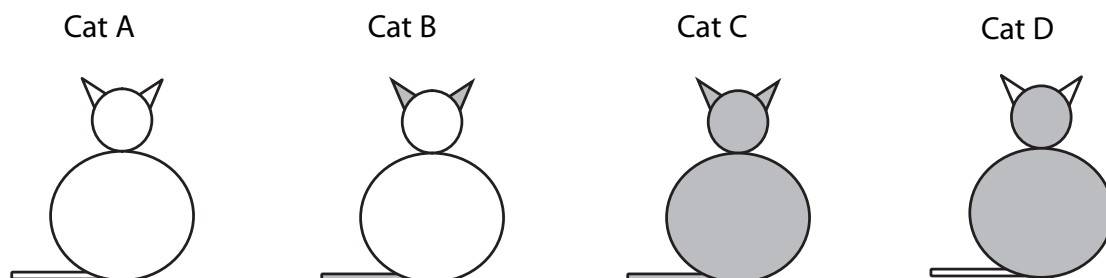
(2)

Animal fur colour is an example of a phenotype. The phenotype of an animal can be the result of an interaction between the and the

- (ii) The enzyme tyrosinase is involved in producing dark coloured fur in cats. Tyrosinase is active at the cat's body temperature.

Some cats have a defective form of tyrosinase. The defective tyrosinase is inactive at normal body temperature, but becomes active in the cooler extremities of cats living in a cold environment.

The diagrams below show four cats with different fur colour distributions. Shaded areas represent dark coloured fur and non-shaded areas represent light coloured fur.



Place a cross in the most appropriate box to select the diagram which represents:

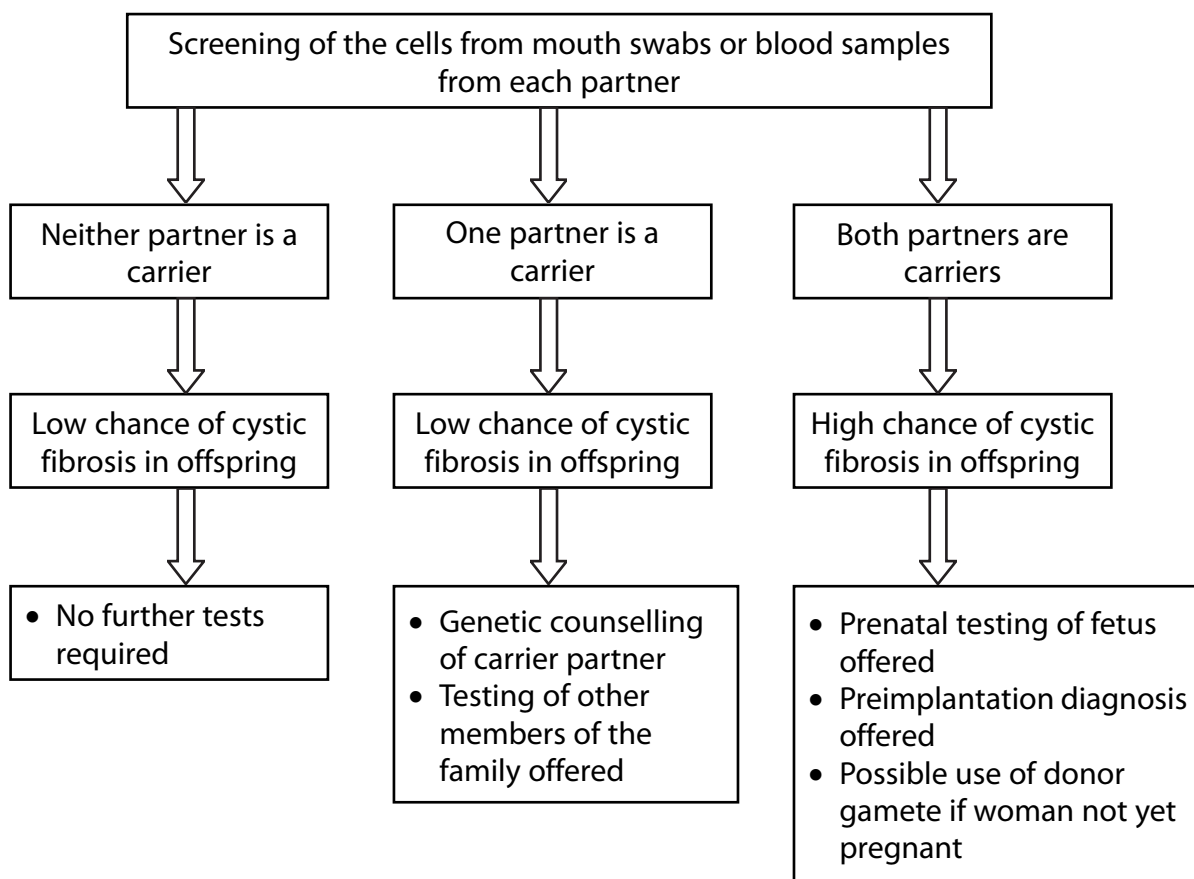
(3)

- | | | | | |
|---|---------------------------------------|---------------------------------------|---------------------------------------|---------------------------------------|
| a cat with normal tyrosinase | <input checked="" type="checkbox"/> A | <input checked="" type="checkbox"/> B | <input checked="" type="checkbox"/> C | <input checked="" type="checkbox"/> D |
| a cat with defective tyrosinase, in a hot country | <input checked="" type="checkbox"/> A | <input checked="" type="checkbox"/> B | <input checked="" type="checkbox"/> C | <input checked="" type="checkbox"/> D |
| a cat in a cold country with defective tyrosinase | <input checked="" type="checkbox"/> A | <input checked="" type="checkbox"/> B | <input checked="" type="checkbox"/> C | <input checked="" type="checkbox"/> D |

(Total for Question 2 = 9 marks)

3 Cystic fibrosis is a life-threatening condition that can affect many different parts of the body. It is a recessive genetic trait. Genetic screening can be used to test for the presence of recessive alleles. A person found to possess a recessive allele is called a carrier.

The diagram below illustrates a risk analysis following the screening for recessive alleles of a couple, who are planning a pregnancy.



(a) Suggest why cells from mouth swabs or blood samples are used rather than gametes.

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(b) Explain why it is necessary to test for several different recessive alleles in the screening for cystic fibrosis.

(2)

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(c) In the risk analysis shown, if neither partner is a carrier then it is considered that the chance of having a child with cystic fibrosis is low. Explain why the probability of having a child with cystic fibrosis is low and **not** zero.

(2)

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(d) In the risk analysis shown, if one of the partners is found to be a carrier then screening for cystic fibrosis may be offered to other family members. Explain why this screening is offered to other family members.

(2)

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(e) Using your knowledge of monohybrid crosses, calculate the probability of having a child with cystic fibrosis if both partners are found to be carriers. Draw a genetic diagram to explain how you calculated this probability.

(5)

Answer

(Total for Question 3 = 13 marks)

4 Albinism is a genetic trait resulting from the inheritance of recessive alleles.

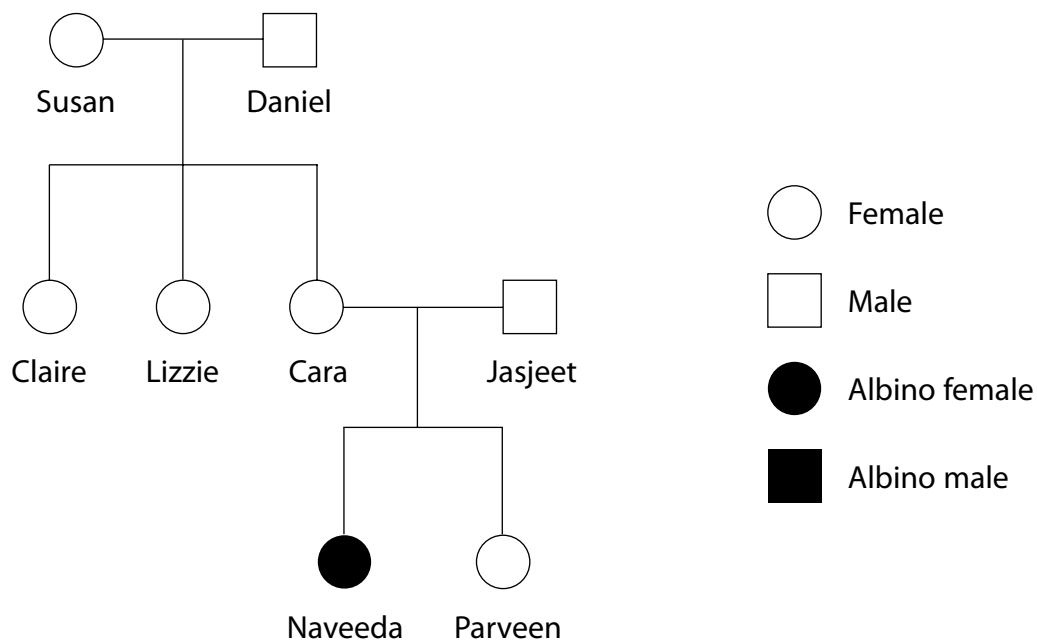
(a) (i) Distinguish between the terms **allele** and **gene**.

(2)

(ii) Explain the meaning of the term **recessive** allele.

(1)

(b) The pedigree diagram below shows the inheritance of albinism in one family.



(i) Naveeda is homozygous. Explain the meaning of the term **homozygous**.

(1)

(ii) Susan is also homozygous. Name the members of this family who are definitely carriers of albinism, giving reasons for your answer.

(4)

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(c) Albinism occurs in a number of different animals, including squirrels as shown in the photograph below.



The incidence of albinism in squirrels is 1 in 100 000 births, which is much lower than the incidence of albinism in humans. Suggest why the incidence of albinism in squirrels is lower than the incidence in humans, giving a reason for your answer.

(2)

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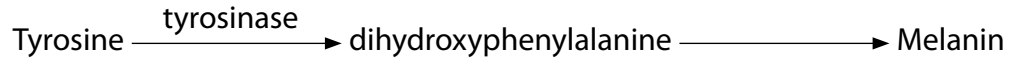
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(d) Individuals with albinism are unable to produce the pigment melanin. This can be due to the absence of the enzyme tyrosinase. The diagram below shows the role of tyrosinase in melanin production.



Explain why melanin cannot be produced in the absence of the enzyme tyrosinase.

(2)

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(Total for Question 4 = 12 marks)