



# Rabbit Fur Colour

A Level



In rabbits, there are two alleles concerned with dark pigment in the fur:

- the dominant allele, B, for black colour
- the recessive allele, b, for brown colour

Two male black rabbits of unknown genotype each mated with a different female brown rabbit.

## Part A Two heterozygous males

What is the expected proportion of brown offspring if **both** male rabbits are heterozygous?

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## Part B One heterozygous male

What is the expected proportion of brown offspring if only **one** male rabbit is heterozygous?

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Adapted with permission from NSAA 2020 Section 1 Q79

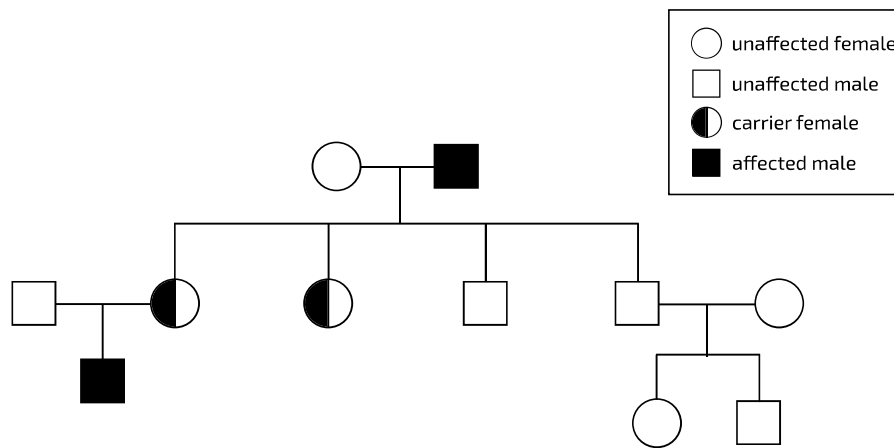
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## Eyesight Problems

A Level



A gene found on the X chromosome in humans has two alleles: dominant and recessive. Individuals who only have recessive alleles have a condition that affects their eyesight. The inheritance of the condition is shown in **Figure 1**.



**Figure 1:** A family tree showing X-linked recessive inheritance. "Unaffected" individuals do not have the condition, "affected" individuals do have the condition, and "carrier" individuals do not have the condition but do carry the recessive allele. The key is shown in the top-right.

### Part A Recessive & dominant alleles

If one living skin cell from each member of this family were analysed, how many recessive alleles (for this gene) would be found?

If one living skin cell from each member of this family were analysed, how many dominant alleles (for this gene) would be found?

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### Part B An affected female and an unaffected male

If an affected female mates with an unaffected male to produce a son, what is the probability that this individual has the condition?

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If an affected female mates with an unaffected male to produce a daughter, what is the probability that this individual has the condition?

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### Part C X-linked recessive inheritance

Which of the following statements are true of X-linked recessive inheritance? Select all that apply.

- ☐ Females cannot have the condition but they can be carriers.
  - ☐ Females are more likely to have the condition than males.
  - ☐ All male offspring of an affected female will have the condition, regardless of the male parent's genotype.
  - ☐ All female offspring of an affected male will have the condition, regardless of the female parent's genotype.
  - ☐ Males cannot be carriers: they either have the condition or they do not.
  - ☐ Males are more likely to have the condition than females.
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Adapted with permission from NSAA 2019 Section 1 Q68

Gameboard:

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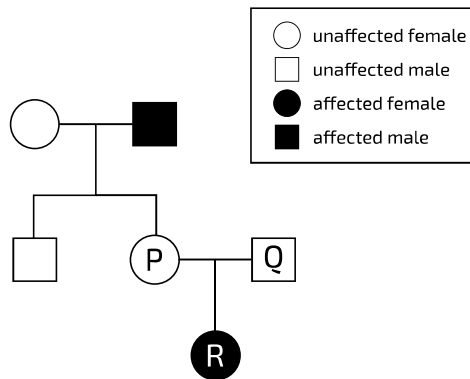
# Dominant and Recessive Conditions

A Level



## Part A A dominant condition

**Figure 1** shows the family tree of a family affected by a dominant genetic condition. All people who carry the mutation show symptoms of the condition.



**Figure 1:** A family tree showing the inheritance of a dominant genetic condition.

Which of the following statements could explain the presence of the dominant condition in female R?

- ☐ The mutation occurs in P.
- ☐ The mutation occurs in R.
- ☐ The mutation occurs in Q's father.

## Part B Cystic fibrosis and sickle cell anaemia

Cystic fibrosis and sickle cell anaemia are both recessive genetic conditions and the genes for these conditions are found on different autosomes.

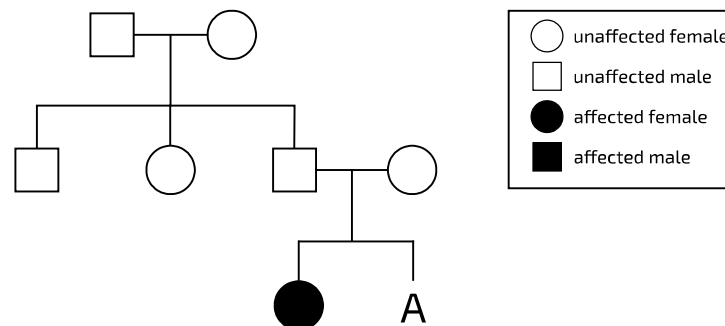
The following statements are true for one set of parents who only have one child:

- Both parents are heterozygous for cystic fibrosis
- One parent is homozygous recessive for sickle cell anaemia
- One parent is heterozygous for sickle cell anaemia

What is the probability of this child having both conditions?

## Part C Pedigree probability

**Figure 2** shows the inheritance of a phenotypic feature caused by a recessive allele. "A" represents an individual of unknown genotype.



**Figure 2:** A family tree showing the inheritance of a recessive genetic condition.

What is the probability that individual A is an unaffected male?

Assume that no mutations occurred to this gene in this family tree.

Adapted with permission from NSAA 2018 Section 1 Q65, NSAA 2021 Section 2 Q41 & NSAA 2021 Section 1 Q74

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## Three Family Trees

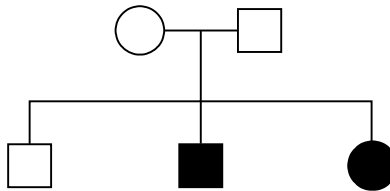
A Level



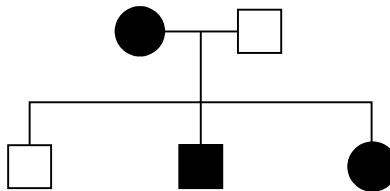
The three family trees below show the inheritance of three different genetic conditions, each controlled by one gene with one dominant and one recessive allele.

Circles represent females and squares represent males.

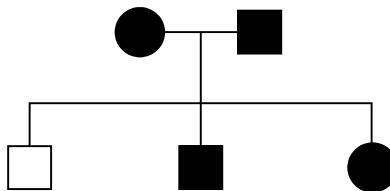
Black circles/squares represent affected individuals and white circles/squares represent unaffected individuals.



Family A



Family B



Family C

In the questions below, assume that no new mutations occur and that the genes are not found on the X chromosome.

## Part A Heterozygous fathers

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In which of the family trees **must** the male parent be heterozygous? Select all that apply.

- ☐ Family A
  - ☐ Family B
  - ☐ Family C
- 

## Part B Dominant conditions

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In which of the family trees **must** the condition be a dominant genetic condition? Select all that apply.

- ☐ Family A
  - ☐ Family B
  - ☐ Family C
- 

## Part C Recessive conditions

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In which of the family trees **must** the condition be a recessive genetic condition? Select all that apply.

- ☐ Family A
  - ☐ Family B
  - ☐ Family C
- 

Adapted with permission from NSAA 2020 Section 1 Q80

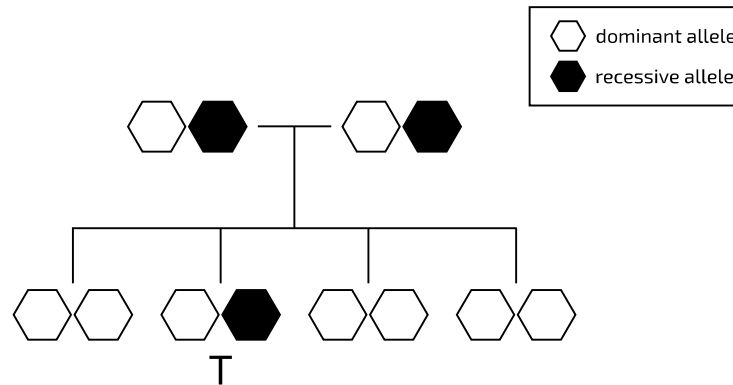
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## An Allele Family Tree

**Figure 1** shows the inheritance of a characteristic controlled by a single gene.



**Figure 1:** A family tree showing the inheritance of a characteristic controlled by a single gene. The genotype of each individual is shown by coloured hexagons (see key at top-right).

Individual T goes on to have a single offspring with an unrelated individual who has a recessive phenotype. The offspring has a dominant phenotype.

What is the ratio of dominant to recessive alleles for all of the individuals in this family (including T's mate and offspring)?

Enter your answer as a ratio in its simplest form (e.g. 10:1).

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## Observed vs Expected Phenotype Ratios

A Level



The chi-squared test can be used to determine whether there is a statistically significant difference between the expected and the observed number of individuals with each phenotype in a population. This test involves calculating a value (the  $\chi^2$  statistic) and comparing this to critical value. If the calculated value is greater than the critical value, then there is a statistically significant difference.

The value required for this test ( $\chi^2$ ) is calculated using the following expression:

$$\frac{(\text{observed} - \text{expected})^2}{\text{expected}} + \dots + \frac{(\text{observed} - \text{expected})^2}{\text{expected}}$$

where each term uses the observed number of individuals and expected number of individuals with each phenotype in turn.

In a monohybrid cross between two individuals that showed the same phenotype, 160 offspring were produced. 36 of these offspring showed a different phenotype to both parents for the same characteristic.

The characteristic is controlled by a single gene with one dominant allele and one recessive allele.

### Part A Chi-squared statistic ( $\chi^2$ )

Calculate  $\chi^2$ . Give your answer to 3 d.p.

(Assumes no mutations occur and that no genotype results in the death of individuals).

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## Part B Statistical significance

Using the significance table in the hint below, find the appropriate critical value of chi-squared at the 5% level of significance.

Give your answer to 3 d.p.

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Is the observed phenotype ratio significantly different from the expected ratio?

☐ yes

☐ no

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