16.2 The roles of genes in determining the phenotype

Name: _____ Class: _____ Date: _____ **Total: 33 marks**

KEY WORDS allele • codominance • sex linkage • albinism • chi-squared test

Objective

Build the skills to answer exam questions on **the roles of genes in determining the phenotype** – genetic terms, **genetic diagrams** (monohybrid, codominance, sex linkage), the **chi-squared test**, and gene→protein→phenotype.

You must be able to:

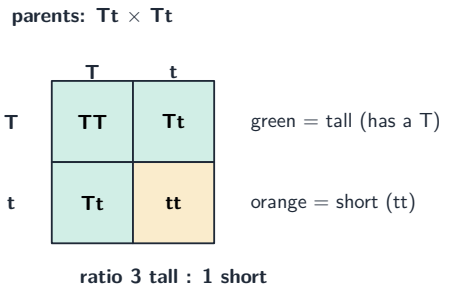
- use the genetic terms exactly and construct Punnett squares for crosses
- predict ratios for monohybrid, codominance and sex-linked crosses
- relate genes to proteins and phenotype (e.g. albinism, sickle cell)

1 Key knowledge and worked examples

The language of genetics

A **gene** is a length of DNA coding for a protein; its position is its **locus**. An **allele** is a version of a gene. **Dominant** shows with one copy; **recessive** needs two. **Homozygous** = same alleles; **heterozygous** = different. **Genotype** = the alleles; **phenotype** = the visible features.

A monohybrid cross

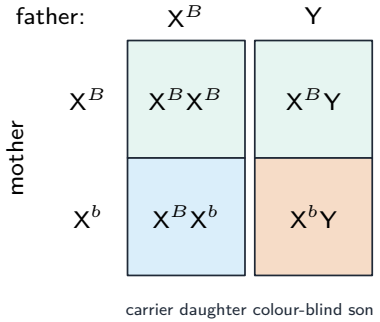


Tt × Tt → 3 tall : 1 short (T dominant)

For Tt × Tt: gametes T and t; offspring TT, Tt, Tt, tt → **3 dominant : 1 recessive**.

Codominance and sex linkage

In **codominance** both alleles show (e.g. red × white flowers → pink). In **sex linkage** the gene is on the X chromosome, so results differ by sex.



X-linked: a carrier mother's sons may be affected while daughters are carriers

Genes, proteins and phenotype

A faulty allele → a faulty protein → a changed phenotype: e.g. the *TYR* gene → tyrosinase → **albinism** if non-functional; *HBB* → haemoglobin → **sickle cell anaemia**.

Reading a cross, and the ratios that are not 3:1

Start every genetics question by writing the **key**, then parental genotypes, gametes, the Punnett square, and finally genotype and phenotype ratios. Each layer carries a mark, so a bare ratio scores one of four or five.

The standard results:

Cross	Phenotype ratio
Aa × Aa (monohybrid)	3 : 1
Aa × aa (test cross)	1 : 1
AaBb × AaBb (dihybrid)	9 : 3 : 3 : 1
AaBb × aabb	1 : 1 : 1 : 1

The ratios that signal something else – this is what separates A-Level from IGCSE:

- **Codominance** : BOTH alleles are expressed in the heterozygote, giving a third phenotype, so $C^RC^W \times C^RC^W$ gives **1 : 2 : 1** with roan or pink in the middle. Write codominant alleles as superscripts on a common letter, never as upper and lower case.
- **Multiple alleles**: three or more alleles exist in the population, though any individual still has two – human ABO blood groups, where I^A and I^B are codominant and both dominant to I^O.
- **Sex linkage** : the gene is on the X chromosome, so a male has only one copy. Write the genotypes as X^HY and X^HX^h, and expect the ratio to **differ between the sexes** – a carrier mother and unaffected father give affected sons but no affected daughters.
- **Autosomal lethal allele**: a 2 : 1 ratio instead of 3 : 1, because the homozygous dominant class dies before birth.
- **Epistasis**: one gene masks another, turning 9 : 3 : 3 : 1 into 9 : 7, 9 : 3 : 4 or 12 : 3 : 1.

The chi-squared test decides whether an observed ratio differs significantly from the expected. Null hypothesis: there is no significant difference between observed and expected, and any difference is due to chance. Degrees of freedom = number of classes − 1. If χ^2 **exceeds** the critical value at $p = 0.05$, **reject** the null hypothesis.

Worked example. In a cross between two roan cattle (C^RC^W), predict the offspring ratio and explain why it is not 3 : 1.

1. Gametes C^R and C^W from each parent.
2. Offspring: C^RC^R, C^RC^W, C^RC^W, C^WC^W.
3. The alleles are **codominant**, so the heterozygote shows both and is roan rather than red.
4. The ratio is therefore **1 red : 2 roan : 1 white**, and the genotype and phenotype ratios are the same – which is the giveaway for codominance.

2 Practice

2.1 Define **homozygous** and **heterozygous**. [2]

2.2 A test cross is used to find an unknown genotype. State the genotype the organism is crossed with. [1]

2.3 State the difference between codominance and incomplete dominance in how the heterozygote appears. [2]

2.4 Explain why a sex-linked condition is far more common in males. [2]

2.5 State the null hypothesis for a chi-squared test on a genetic cross. [2]

3 Exam-style questions

3.1 In a cross Tt × tt, what proportion of the offspring are expected to show the recessive phenotype? [1]

☐ **A** 0
B 1/4
C 1/2
D 3/4

3.2 Two pink flowers (codominant red R and white W, genotype RW) are crossed. The expected ratio of red : pink : white is: [1]

☐ **A** 3 : 0 : 1
B 1 : 2 : 1
C 1 : 1 : 0
D 9 : 3 : 4

3.3 Red-green colour blindness is X-linked recessive. A carrier mother (X^BX^b) has children with a normal father (X^BY).

(a) Draw a genetic diagram (Punnett square) for this cross. [3]

(b) State the probability that a son is colour-blind. [1]

3.4 Explain how a faulty *TYR* allele leads to albinism. [2]

3.5 In cats, the gene for coat colour is carried on the X chromosome. The allele X^O gives orange fur and X^B gives black; they are codominant, and a cat with both is tortoiseshell.

(a) Write the genotypes of an orange male, a black female and a tortoiseshell female. [3]

(b) A tortoiseshell female is crossed with an orange male. Draw the genetic diagram and give the genotypes and phenotypes of the offspring. [5]

(c) Explain why tortoiseshell males are extremely rare. [2]

(d) In a different cross, 96 offspring were obtained: 54 orange and 42 black. The expected ratio was $1:1$. Calculate the χ^2 value. [3]

(e) The critical value at $p = 0.05$ with 1 degree of freedom is 3.84. State and explain the conclusion. [3]

Solutions

Each answer shows the steps, then the **result**.

2.1

- homozygous – the two alleles of a gene are the same; heterozygous – the two alleles are different

2.2

- the homozygous recessive

2.3

- In codominance both alleles are fully expressed, so the heterozygote shows both phenotypes together, such as roan; in incomplete dominance the heterozygote shows an intermediate blend, such as pink

2.4

- The gene is on the X chromosome and a male has only one X, so a single recessive allele is expressed with no second allele to mask it, whereas a female would need two

2.5

- There is no significant difference between the observed and the expected numbers, and any difference is due to chance

3.1 C

- Tt × tt
- Tt, Tt, tt, tt
- half show the recessive phenotype

3.2 B

- RW × RW
- RR (red) : RW (pink) : RW (pink) : WW (white) = 1 : 2 : 1

3.3

(a)

- parents $X^BX^b \times X^BY$; gametes X^B, X^b and X^B, Y; offspring $X^BX^B, X^BX^b, X^BY, X^bY$ – normal daughter, carrier daughter, normal son, colour-blind son

(b)

- 1/2 (of the sons)

3.4

- the faulty *TYR* allele codes for a non-functional tyrosinase enzyme; so no melanin pigment is made, giving albinism

3.5

(a)

- Orange male X^OY; black female X^BX^B; tortoiseshell female X^OX^B

(b)

- Parents $X^OX^B \times X^OY$; gametes X^O, X^B and X^O, Y; offspring X^OX^O, X^OX^B, X^OY, X^BY; phenotypes orange female, tortoiseshell female, orange male, black male; ratio 1 : 1 : 1 : 1

(c)

- A tortoiseshell cat needs both X^O and X^B, so it needs two X chromosomes; a male is normally XY, so only an individual with an abnormal XXY karyotype can be a tortoiseshell male

(d)

- Expected is 48 of each; $\chi^2 = \dfrac{(54-48)^2}{48} + \dfrac{(42-48)^2}{48} = \dfrac{36}{48} + \dfrac{36}{48} = 1.5$

(e)

- 1.5 is less than the critical value of 3.84; so the null hypothesis is retained; there is no significant difference between the observed and expected numbers, and the difference is due to chance