

Exercise walk-through

16.2

The roles of genes in determining the phenotype

A-Level Biology

From genes to phenotype

faulty **tyrosinase** →
albinism

faulty **haemoglobin** →
sickle cell

faulty factor VIII →
haemophilia

faulty huntingtin → **Huntington's**

A gene codes for a protein, and the protein shapes the **phenotype** – so a faulty allele gives a faulty protein.

Gibberellin and stem height

In pea plants, the dominant allele *Le* codes for a working enzyme that makes **gibberellin**, so the plant grows tall by stem **elongation**.

The recessive allele *le* codes for a broken enzyme, so little gibberellin is made and the plant is short.

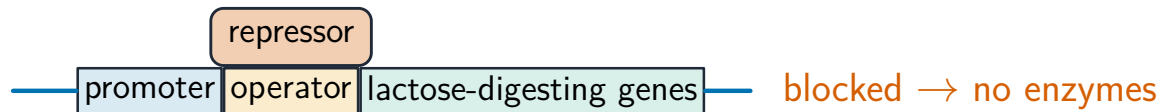
Gene control: the lac operon

The **lac operon** controls lactose digestion:

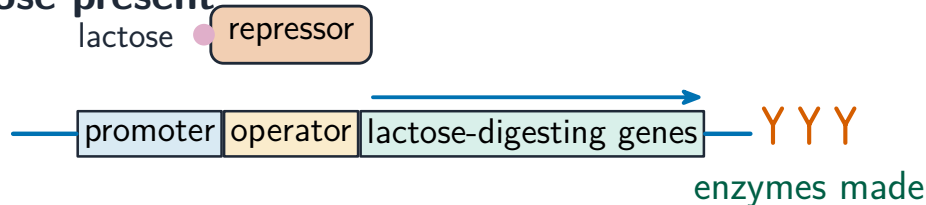
- no lactose → a **repressor** blocks the genes
- lactose pulls the repressor off → enzymes made (**inducible**)

Eukaryotes use **transcription factors**.

no lactose



lactose present



Switching a gene on when it is needed

The operon

In a **prokaryote**
operon

such as a bacterium, a group of genes – the **lac**
– controls the digestion of lactose.

Off by default

A repressor protein binds the operator, so RNA polymerase cannot transcribe the genes. No lactose, no enzymes, no waste.

Lactose switches it on

Lactose binds the repressor and changes its shape, so it lets go, and transcription runs.

Two kinds of enzyme

An **inducible enzyme**
zyme

is made only when it is needed; a **repressible enzyme**
is normally made and can be switched off.

The cell is not deciding anything. A molecule changes a protein's shape, and the shape change is the decision.

The chi-squared test, and how gibberellin switches genes on

the chi-squared test

compares the numbers you actually counted (**observed**, O) with the numbers a hypothesis predicts (**expected**, E)

the statistic

$$\chi^2 = \sum \frac{(O - E)^2}{E}$$

the degrees of freedom

one fewer than the number of categories

the conclusion

if χ^2 exceeds the critical value at $p = 0.05$, the difference is **unlikely to be chance**

gibberellin

causes the breakdown of **DELLA** protein repressors

the effect

with the repressors gone, the genes they were blocking are transcribed – so the hormone acts by **removing a brake**

An **inducible enzyme** is made only when needed. Both gibberellin and the lac operon work by **lifting repression**, not by adding a signal to switch on.

Exercises 2.1-2.3

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]

Exercises 2.1-2.3

2.1 Define **homozygous** and **heterozygous**.

[2]

2.1

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

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

[1]

2.2

- the homozygous recessive

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

[2]

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

Exercises 2.4-2.5

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]

Exercises 2.4-2.5

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

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

2.5

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

Exercise 3.1

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

☐

A 0

B $1/4$

C $1/2$

D $3/4$

Exercise 3.1

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

☐

A 0

B $1/4$

C $1/2$

D $3/4$

3.1 C

- $Tt \times tt$
- Tt, Tt, tt, tt
- half show the recessive phenotype

Exercise 3.2

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

Exercise 3.2

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

3.2 B

- $RW \times RW$
- RR (red) : RW (pink) : RW (pink) : WW (white) = 1 : 2 : 1

Exercise 3.3 (a)

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

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

[3]

Exercise 3.3 (a)

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

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

(a)

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

Exercise 3.3 (b)

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

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

[1]

Exercise 3.3 (b)

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

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

(b)

- 1/2 (of the sons)

Exercise 3.4

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

[2]

Exercise 3.4

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

3.4

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

Exercise 3.5 (a)

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]

Exercise 3.5 (a)

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]

(a)

- Orange male $X^O Y$; black female $X^B X^B$; tortoiseshell female $X^O X^B$

Exercise 3.5 (b)

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.

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

Exercise 3.5 (b)

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.

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

(b)

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

Exercise 3.5 (c)

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.

(c) Explain why tortoiseshell males are extremely rare.

[2]

Exercise 3.5 (c)

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.

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

(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

Exercise 3.5 (d)

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.

(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]

Exercise 3.5 (d)

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.

(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]

(d)

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

Exercise 3.5 (e)

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.

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

Exercise 3.5 (e)

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.

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

(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