Week 45

A-Level Biology

Homework bundle for this week

本周作业合集

exercises.lol

Contents 目录

A-Level Biology Name: _____ Score: _____

1. Why must gametes be haploid?
   - ☐ so the chromosome number does not double at every generation
   - ☐ so they can move faster
   - ☐ so they have more chromosomes
   - ☐ so they can photosynthesise
2. An allele is:
   - ☐ one version of a gene
   - ☐ the position of a gene on a chromosome
   - ☐ a whole chromosome
   - ☐ a type of protein
3. A Punnett square is used to predict the offspring genotypes and ratios of a genetic cross.

   ☐ True ☐ False
4. How does a faulty allele change the phenotype?
   - ☐ it makes a faulty protein, which changes the feature the protein controls
   - ☐ it adds a new chromosome
   - ☐ it has no effect on the protein
   - ☐ it changes the number of genes
5. A regulatory gene:
   - ☐ controls whether other genes are switched on
   - ☐ always codes for an enzyme
   - ☐ is never expressed
   - ☐ makes the cell wall
6. Meiosis halves the chromosome number, producing _____ gametes (one set of chromosomes each).

   _____
7. A recessive allele:
   - ☐ only shows its effect when two copies are present
   - ☐ shows its effect with just one copy

☐ always masks the dominant allele

☐ is never inherited

8. A faulty allele changes the phenotype because it codes for a faulty protein.

   ☐ True ☐ False

9. An enzyme that is made only when it is needed (like the lac enzymes) is described as _____.

   _____

10. The observable features of an organism (what you can actually see) are its _____.

    _____

16. Inheritance

Answers · 答案

A-Level Biology

1. **so the chromosome number does not double at every generation**

   If gametes were diploid, fertilisation would double the chromosome number each generation; meiosis halves it.

2. **one version of a gene**

   An allele is a version of a gene; its position on the chromosome is the locus.

3. **True**

   It combines every possible pairing of the parents' gametes.

4. **it makes a faulty protein, which changes the feature the protein controls**

   A gene codes for a protein; a faulty allele makes a faulty protein, altering the phenotype.

5. **controls whether other genes are switched on**

   Structural genes code for proteins like enzymes; regulatory genes control whether other genes are switched on.

6. **haploid**

   Two haploid gametes fuse at fertilisation to restore the diploid number.

7. **only shows its effect when two copies are present**

   A recessive allele is only expressed when homozygous; a dominant allele shows with a single copy.

8. **True**

   The DNA sequence sets the protein's shape; change the sequence and you change (or break) the protein.

9. **inducible**

   Inducible enzymes are produced only when required, which avoids wasting resources.

10. **phenotype**

    Phenotype = what shows; genotype = the alleles present (which, with the environment, decide the phenotype).

A-LEVEL BIOLOGY • EXERCISE SHEET

16.1 Passage of information from parents to offspring

Name: _____ Class: _____ Date: _____ **Total: 25 marks**

KEY WORDS gamete 配子 • meiosis 减数分裂 • haploid 单倍体 • diploid 二倍体 • crossing over 交叉互换

Objective

Build the skills to answer exam questions on the **passage of information from parents to offspring** — **meiosis** 减数分裂, and how **crossing over** 交叉互换, **independent assortment** 自由组合 and fertilisation create variation.

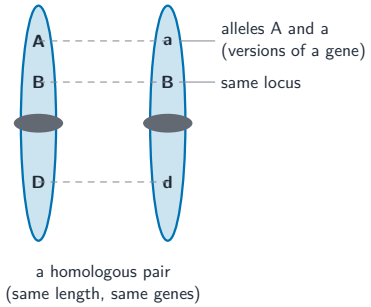
You must be able to:

- explain haploid and diploid, homologous pairs, and the need for a reduction division
- describe the behaviour of chromosomes during meiosis and identify its stages
- explain how meiosis and fertilisation produce genetically different individuals

1 Worked examples

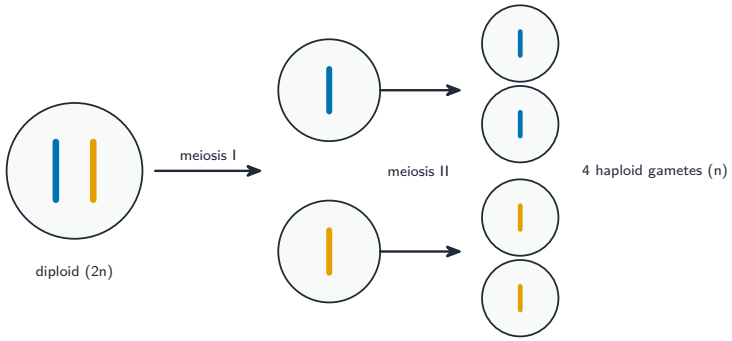
Haploid, diploid and homologous pairs

A **diploid** 二倍体 cell (2n) has two sets of chromosomes; a **haploid** 单倍体 cell (n) has one. **Homologous chromosomes** 同源染色体 are a pair of the same length carrying the same genes (alleles may differ).



A homologous pair carries the same genes at the same loci; the alleles may differ

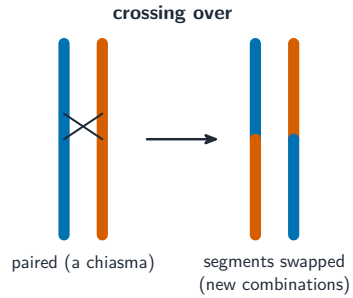
Meiosis halves the chromosome number



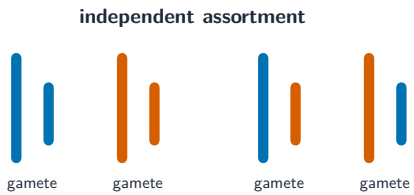
Meiosis I separates the homologous pair; meiosis II separates the chromatids → four haploid gametes

Gametes must be haploid, or the chromosome number would double each generation. Meiosis has two divisions (meiosis I and II).

How meiosis makes variation



Crossing over swaps matching segments, making new allele combinations



Independent assortment: pairs line up randomly, so gametes get many different mixes

Then **random fertilisation** lets any gamete fuse with any other, so every new individual is genetically different.

2 Practice

2.1 State the difference between a haploid and a diploid cell. [2]

2.2 Name the two processes during meiosis that make gametes genetically different. [2]

3 Exam-style questions

3.1 Why must gametes be produced by a reduction division? [1]

- **A** to make the gametes larger
- **B** to keep the chromosome number constant from generation to generation
- **C** to copy the DNA
- **D** to produce identical cells

3.2 During which division of meiosis are homologous chromosomes separated? [1]

- **A** meiosis I
- **B** meiosis II
- **C** mitosis
- **D** fertilisation

3.3 Explain how crossing over and independent assortment each increase genetic variation. [4]

3.4 An organism has a haploid number of $n = 4$.

(a) Calculate the number of different chromosome combinations possible in its gametes from independent assortment alone (2^n). [2]

(b) Explain why the actual number of genetically different gametes is even larger than this. [1]

3.5 A diploid animal has $2n = 6$.

(a) Describe **three** events in meiosis that produce genetic variation, naming the stage at which each occurs. [6]

(b) Calculate the number of genetically different gametes this animal could produce by independent assortment alone, showing your working. [2]

(c) Explain why the number of possible offspring is far larger than your answer to (b). [2]

(d) State **two** differences between the products of meiosis and the products of mitosis. [2]

Solutions

2.1 haploid has one set of chromosomes (n); diploid has two sets (2n).

2.2 crossing over; independent assortment (random orientation).

3.1 B. Halving the number means fertilisation restores the diploid number, keeping it constant.

3.2 A. Homologous chromosomes are separated in meiosis I.

3.3 crossing over: homologous chromosomes swap matching segments at chiasmata, making new combinations of alleles; independent assortment: the homologous pairs line up in a random order at the equator, so gametes receive different mixes of maternal and paternal chromosomes.

3.4 (a) $2^4 = 16$.
(b) crossing over also creates new allele combinations, adding to the variety.

3.5 (a) **Crossing over** in prophase I — non-sister chromatids of a bivalent exchange lengths of DNA at a chiasma, producing new combinations of alleles on one chromosome. **Independent assortment** in metaphase I — each bivalent lines up on the equator independently of the others, so either homologue may go to either pole. **Random assortment of chromatids** in metaphase II — the sister chromatids, no longer identical after crossing over, separate independently.
(b) 2^n where n is the haploid number, here 3; $2^3 = 8$ different gametes.
(c) Crossing over creates chromatids that are not simply one homologue or the other, so the real number of gamete types is far greater than 8; and fertilisation is random — any one of the male gametes may fuse with any one of the female gametes, multiplying the possibilities.
(d) Meiosis gives four haploid cells, mitosis two diploid cells; meiotic products are all genetically different from each other and from the parent, mitotic products are genetically identical to it.

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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..... [2]

(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.

..... [1]

(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.

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..... [2]

[Total: 9]

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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..... [4]

(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.

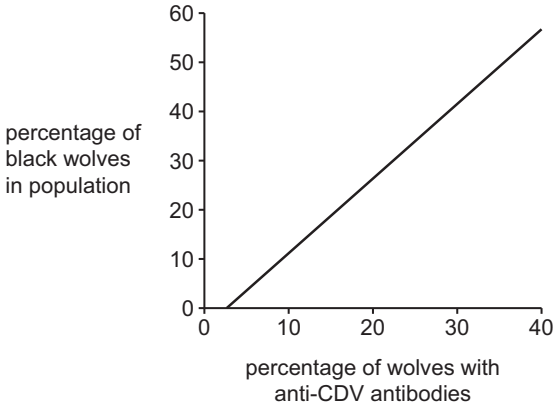

Fig. 4.1

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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..... [4]

[Total: 11]

5 Metachromatic leukodystrophy (MLD) is a genetic disease that affects the nervous system.

MLD is caused by mutations in the *ARSA* gene located on chromosome 22. The *ARSA* gene, which is 3150 base pairs (bp) in length, includes 8 exons and is shown in Fig. 5.1.

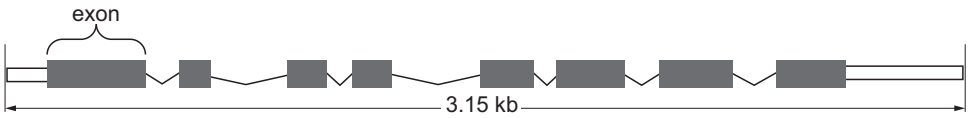

Fig. 5.1

A genetic test using DNA sequencing is available to identify mutations associated with MLD in the *ARSA* gene. The sequencing method can only work if a DNA fragment size is less than 1000 bp.

The test involves a number of different stages:

- Genomic DNA is extracted from a blood sample.
- Polymerase chain reaction (PCR) using 5 pairs of primers selects 5 different DNA fragments of the *ARSA* gene.
- Gel electrophoresis is carried out on the DNA fragments.
- The DNA fragments are sequenced and analysed for mutations.

Table 5.1 shows the length of the DNA fragments and the exons present in each DNA fragment.

Table 5.1

fragment number	DNA fragment length / bp	exons in DNA fragment
1	405	1
2	737	1-2
3	706	2-4
4	860	5-7
5	916	7-8

(a) Genomic DNA and primers are added to the PCR machine.

Name **two** other substances that are added to the PCR machine, **and** explain why each substance is added.

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..... [4]

(b) Gel electrophoresis is carried out on the 5 different DNA fragments produced as a result of PCR. Each DNA fragment is put into a loading well on the gel.

The final lane on the electrophoresis gel contains a DNA ladder of known lengths of DNA.

Fig. 5.2 shows the results of the gel electrophoresis.

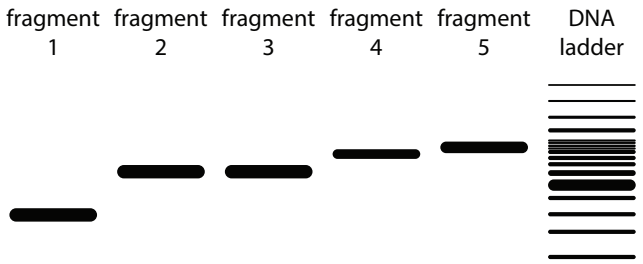

Fig. 5.2

(i) Suggest the role of the DNA ladder.

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..... [2]

(ii) In the genetic test for MLD, PCR and gel electrophoresis are carried out before DNA sequencing.

Suggest **and** explain reasons why PCR and gel electrophoresis are carried out before DNA sequencing.

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..... [3]

(c) Calculate the number of copies of DNA that are obtained if PCR is run for 35 cycles.

Assume that there is only 1 copy of DNA at the start of PCR.

Give your answer in standard form to **two** significant figures.

number of copies .....
[1]

(d) MLD is a rare autosomal recessive disease. It is estimated that there is one case of MLD in every 40 000 births worldwide. Children with MLD have a reduced life expectancy.

- MLD is a degenerative disease that causes severe disability.
- Before 2022, there was no known cure for MLD and the only treatment for symptoms was to provide pain relief.

A new gene therapy treatment known as Libmeldy® became available in a number of countries in 2022. The treatment must start before symptoms are present. Although the treatment may provide a cure for MLD, a single dose is extremely expensive.

Blood samples from newborn babies are screened for a number of rare genetic diseases (neonatal screening) in most countries of the world, but none include screening for MLD.

Discuss the social and ethical considerations of including screening for MLD when carrying out neonatal screening in a country where gene therapy for MLD is available.

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..... [4]

[Total: 14]

2 **(a)** Meiosis and cytokinesis occur in the male reproductive organs (anthers) of plants to make pollen grains. Cells which carry out meiosis are known as pollen mother cells.

Fig. 2.1 shows five stages of meiosis in a pollen mother cell.

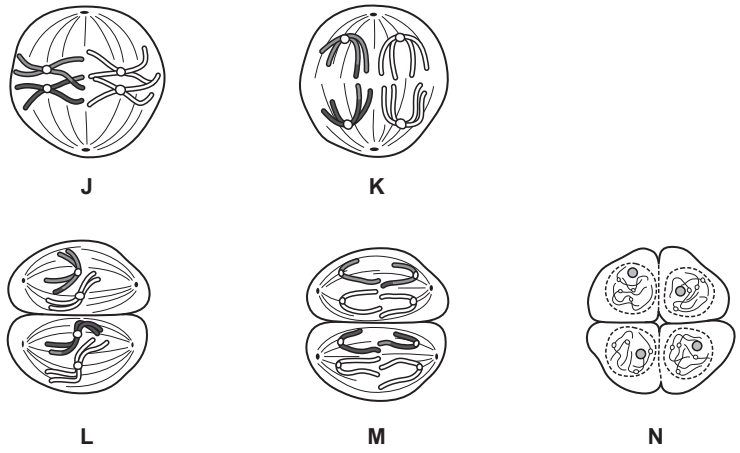

Fig. 2.1

Use the letters **J**–**N** in Fig. 2.1 to state **all** the stages that show:

anaphase .....

cells containing pairs of homologous chromosomes .....

crossing over .....

haploid cells .....

[4]

(b) Maize plants have male and female reproductive organs on the same plant. The male anthers are located on structures known as tassels. When selective breeding is carried out to create an F1 hybrid, the tassels are removed.

Suggest why the tassels are removed when selective breeding is carried out to create an F1 hybrid.

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..... [2]

(c) Outline how selective breeding is used to produce vigorous, uniform varieties of maize.

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..... [4]

[Total: 10]

5 The fruit fly, *Drosophila melanogaster*, feeds on sugars found in damaged fruits.

A fruit fly with normal features is described as wild type. It has a grey body and its wings are longer than its abdomen. The genes for body colour and wing length are located on different chromosomes.

A fruit fly with mutations in these two genes has a black body and short wings.

Fig. 5.1 shows a wild type fruit fly and a mutant fruit fly.

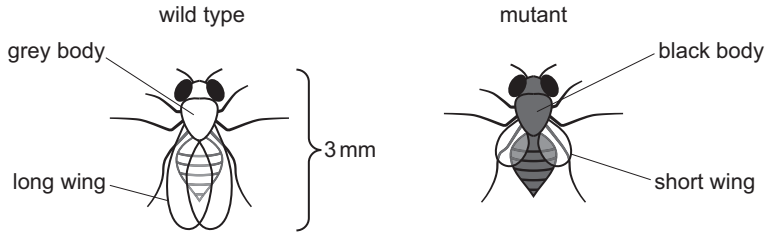

Fig. 5.1

(a) Fruit flies were first used for genetic crosses by Thomas Morgan in 1908. They are one of the most studied animals in current genetic research.

- Male fruit flies are easily distinguished from female fruit flies.
- Fruit flies have a short life cycle and a female can lay hundreds of eggs in a few days.
- Some genes for development and cell signalling in fruit flies are similar to those of humans.

Suggest why fruit flies are still used in genetic crosses.

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..... [3]

(b) When a wild type fruit fly was crossed with a mutant fruit fly with a black body and short wings, all the F1 offspring had grey bodies and long wings.

Using appropriate symbols, complete Fig. 5.2 to show the expected results of a cross between two of these **F1** fruit flies.

symbols:

F1 phenotypes: grey body long wing × grey body long wing

F1 genotypes:

Punnett square

offspring phenotypes:

phenotypic ratio .....

Fig. 5.2

[6]

(c) Describe how you would determine the genotype of an F2 fruit fly with a grey body and long wings.

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..... [3]

[Total: 12]

A-LEVEL BIOLOGY • EXERCISE SHEET

16.2 The roles of genes in determining the phenotype

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

KEY WORDS allele 等位基因 • codominance 共显性 • sex linkage 伴性遗传 • albinism 白化病 • chi-squared test 卡方检验

Objective

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

You must be able to:

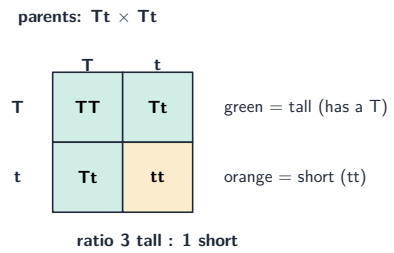
- use the genetic terms exactly and construct Punnett squares for crosses
- predict ratios for monohybrid, codominance and sex-linked crosses
- relate genes to proteins and phenotype (e.g. albinism, sickle cell)

1 Worked examples

■ The language of genetics

A **gene** is a length of DNA coding for a protein; its position is its **locus** 基因座. An **allele** 等位基因 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

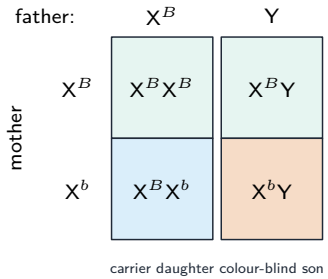


Tt × Tt → 3 tall : 1 short (T dominant)

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

■ Codominance and sex linkage

In **codominance** 共显性 both alleles show (e.g. red × white flowers → pink). In **sex linkage** 伴性遗传 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** 白化病 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** 共显性: 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** 伴性遗传: 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

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; $\chi^2 = \frac{(54-48)^2}{48} + \frac{(42-48)^2}{48} = \frac{36}{48} + \frac{36}{48} = 1.5$.
(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.

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.

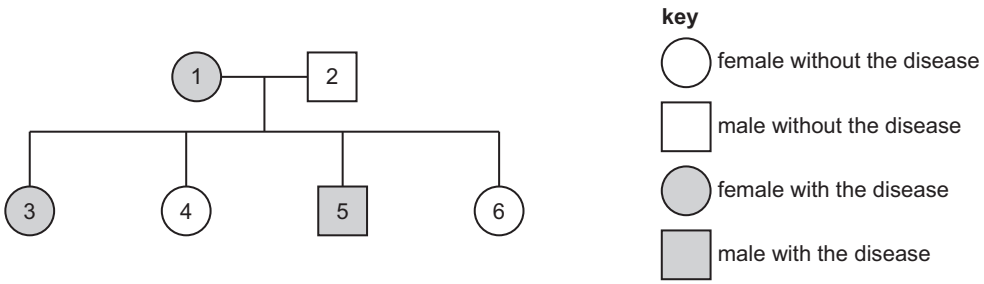


Fig. 3.1

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.

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(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.

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..... [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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..... [1]

(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.

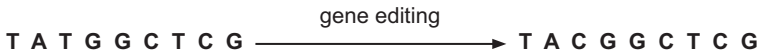

Fig. 3.2

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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..... [3]

A-LEVEL BIOLOGY • EXERCISE SHEET

16.3 Gene control

Name: _____ Class: _____ Date: _____ **Total: 23 marks**

KEY WORDS lac operon 乳糖操纵子 • regulatory gene 调节基因 • structural gene 结构基因 • transcription 转录 • transcription factor 转录因子

Objective

Build the skills to answer exam questions on **gene control** — **structural and regulatory genes** 结构/调节基因, **inducible and repressible enzymes** 可诱导/可抑制酶, the **lac operon** 乳糖操纵子, and **transcription factors** 转录因子.

You must be able to:

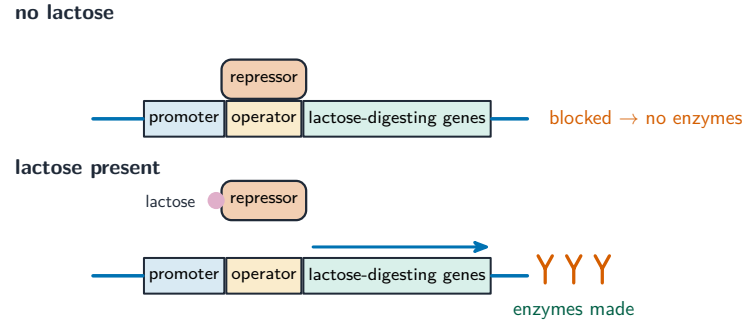
- distinguish structural and regulatory genes, and inducible and repressible enzymes
- explain the control of protein production by the lac operon
- state the role of transcription factors and explain gibberellin's action via DELLA proteins

1 Worked examples

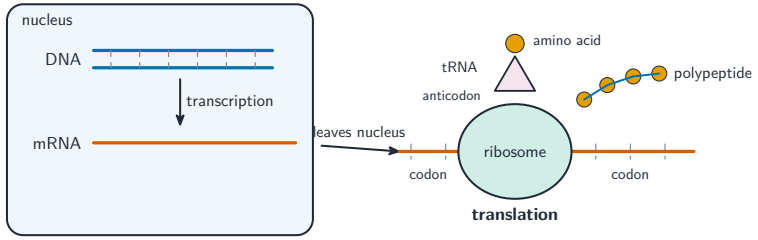
Genes and enzymes

A **structural gene** codes for a useful protein (e.g. an enzyme); a **regulatory gene** controls whether other genes are switched on. An **inducible enzyme** is made only when needed; a **repressible enzyme** is normally made but can be switched off.

The lac operon



A repressor blocks the genes until lactose pulls it off — so the enzymes are inducible



Gene control acts on transcription and translation

- **no lactose**: a **repressor** 阻遏物 protein binds the operator and blocks **transcription** 转录 — no enzymes made.
- **lactose present**: lactose binds the repressor and pulls it off; transcription occurs and the lactose-digesting enzymes are made (they are **inducible**).

■ Transcription factors and gibberellin

In eukaryotes, **transcription factors** are proteins that bind DNA and raise or lower the rate of transcription. **Gibberellin** 赤霉素 switches genes on by causing the breakdown of **DELLA** protein repressors, which normally block the factors that promote transcription.

2 Practice

2.1 State the difference between a structural gene and a regulatory gene. [2]

2.2 State what is meant by an inducible enzyme. [1]

3 Exam-style questions

3.1 In the lac operon, when **no** lactose is present, the repressor protein: [1]

- **A** is broken down
- **B** binds the operator and blocks transcription
- **C** is transcribed into mRNA
- **D** binds the lactose-digesting enzymes

3.2 What is the role of a transcription factor in a eukaryotic cell? [1]

- **A** to replicate DNA
- **B** to bind DNA and control the rate of transcription
- **C** to translate mRNA
- **D** to break down glucose

3.3 Explain how the lac operon allows a bacterium to make lactose-digesting enzymes only when lactose is present. [4]

3.4 Explain how gibberellin switches on genes by acting on DELLA proteins. [2]

3.5 In *Escherichia coli* the *lac* operon controls the enzymes that break down lactose.

(a) Distinguish between a structural gene and a regulatory gene, using the *lac* operon as your example. [3]

(b) Describe what happens at the operon when **no** lactose is present. [3]

(c) Describe what happens when lactose **is** present, and explain why the enzymes are described as inducible. [4]

(d) Explain the advantage to the bacterium of controlling these genes in this way. [2]

Solutions

2.1 a structural gene codes for a (useful) protein such as an enzyme; a regulatory gene controls whether other genes are switched on.

2.2 an enzyme that is made only when it is needed (e.g. when its substrate is present).

3.1 B. With no lactose, the repressor binds the operator and blocks transcription.

3.2 B. A transcription factor binds DNA and increases or decreases the rate of transcription.

3.3 when no lactose is present, the repressor binds the operator and blocks transcription, so no enzymes are made; when lactose is present, it binds the repressor and removes it from the operator; transcription can now occur, so the lactose-digesting enzymes are made.

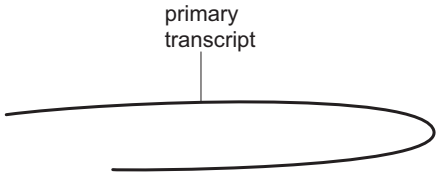
3.4 gibberellin causes the breakdown of DELLA protein repressors; these normally block the transcription factors, so removing them lets the genes be transcribed/expressed.

3.5 (a) A structural gene codes for a protein used by the cell — here *lacZ*, coding for β-galactosidase; a regulatory gene codes for a protein that controls the transcription of other genes — here *lacI*, coding for the repressor.
(b) The regulatory gene is transcribed and the **repressor** protein is made; the repressor binds to the operator region; this blocks RNA polymerase from moving along the structural genes, so no mRNA and no enzyme are made.
(c) Lactose acts as the **inducer**: it binds to the repressor and changes its shape so that it can no longer bind to the operator; RNA polymerase is then free to transcribe the structural genes and the enzymes are made. They are *inducible* because they are produced only when their substrate is present to switch them on.
(d) Making enzymes costs amino acids and ATP, so producing them when there is no lactose to digest would be wasteful; the bacterium makes them only when they can be used, which gives it a selective advantage.

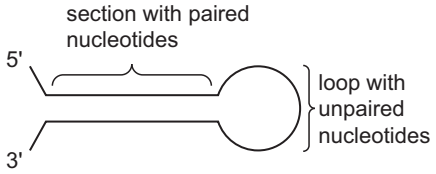
3 Small interfering RNA (siRNA) is a short length of double-stranded RNA (dsRNA), approximately 21 to 25 nucleotides long. siRNA helps to regulate protein synthesis in cells.

(a) Fig. 3.1 is an outline summary of one way in which a primary transcript can be processed to produce a molecule of siRNA. The transcript does **not** code for a sequence of amino acids.

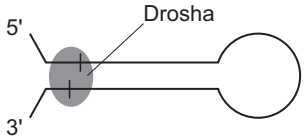
step 1: a primary transcript is synthesised from one strand of DNA



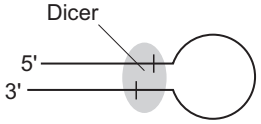
step 2: the primary transcript loops and a section of the RNA becomes double stranded



step 3: the enzyme Drosha attaches and cleaves (cuts) the RNA to produce a shorter length



step 4: in the cytoplasm, the enzyme Dicer attaches and cleaves the RNA



step 5: siRNA, which is double stranded and has 2 unpaired nucleotides at each 3' end, is produced

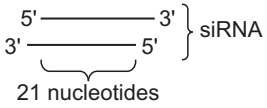


Fig. 3.1

(i) In **step 2** in Fig. 3.1, the single-stranded primary transcript loops and forms a section where dsRNA is present.

Explain how it is possible for the double-stranded section of RNA to be held together in **step 2** and maintained this way in **steps 3**, **4** and **5**.

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..... [2]

(ii) After **step 3**, the shorter dsRNA produced by the action of Drosha is transported to the cytoplasm, where it is cleaved further by Dicer to produce ds siRNA.

Suggest why two different enzymes, Drosha and Dicer, are needed to cut dsRNA into shorter lengths.

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..... [2]

(b) From 2018, siRNA has been used as a therapeutic drug to treat a number of diseases.

The presence of molecules of siRNA in the cytoplasm can result in the cleavage of messenger RNA (mRNA) molecules coding for a protein involved in the disease. This prevents the synthesis of the protein.

Describe the differences between a molecule of mRNA and a molecule of siRNA, such as the siRNA shown in **step 5** in Fig. 3.1.

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..... [2]

(c) Fig. 3.2 summarises how mRNA coding for a protein involved in disease can be targeted and cleaved by siRNA.

presence of siRNA activates a protein, AGO2, which binds siRNA and other proteins to form a complex known as RISC

passenger strand is separated from RISC

RISC uses guide strand to target and bind mRNA for cleavage

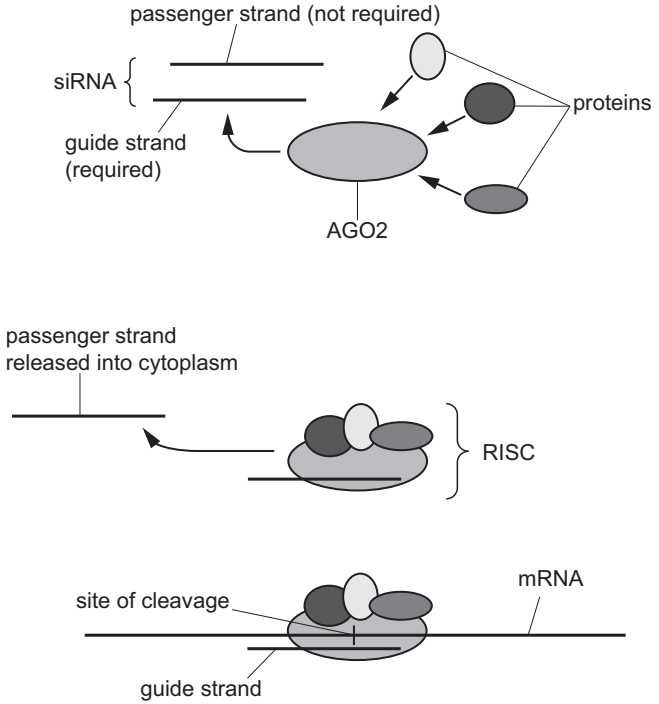

Fig. 3.2

(i) With reference to Fig. 3.2, state why the passenger strand needs to be separated and released from the RISC.

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..... [1]

(ii) The aim of siRNA therapy is to prevent or decrease the synthesis of a protein involved in the disease being treated.

A target mRNA molecule can be cleaved in a different location by a RISC with a different siRNA.

Suggest how cleaving mRNA in different locations will have different effects on protein synthesis **and** explain how these different effects can result in a lack of functioning protein.

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[Total: 10]

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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..... [2]

(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.

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..... [2]

[Total: 9]