Week 48

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. Most features, such as height, are caused by:
   - ☐ both genetic and environmental factors
   - ☐ genetic factors only
   - ☐ environmental factors only
   - ☐ neither genes nor environment
2. Natural selection acts on the variation that already exists in a population.

   ☐ True ☐ False
3. A change in the allele frequencies of a population over generations is called _____.

   _____
4. Selective breeding (artificial selection) differs from natural selection because:
   - ☐ humans choose which organisms breed, rather than nature
   - ☐ no alleles are passed on
   - ☐ it never changes the population
   - ☐ it only happens in bacteria
5. Evolution is best described as:
   - ☐ the gene pool of a population changing over generations, forming new species
   - ☐ an individual changing during its life
   - ☐ the loss of all genetic variation
   - ☐ a single mutation
6. Continuous variation is:
   - ☐ a smooth range controlled by many genes plus the environment
   - ☐ clear separate groups controlled by one gene
   - ☐ caused only by the environment
   - ☐ the same in every individual
7. In natural selection, which individuals are most likely to pass on their alleles?
   - ☐ those best adapted, which survive and reproduce
   - ☐ the largest ones only

☐ a random selection

☐ those that produce the fewest offspring

8. When humans choose which organisms breed to pass on useful features, it is called _____ selection.

_____

9. Speciation is the formation of a new species, when two populations can no longer interbreed.

☐ True ☐ False

10. Repeated artificial selection from a few individuals can reduce the gene pool and cause inbreeding problems.

☐ True ☐ False

17. Selection and evolution

Answers · 答案

A-Level Biology

1. **both genetic and environmental factors**

   Height depends on the alleles inherited and on diet/lifestyle — a combination of genes and environment.

2. **True**

   It cannot create variation; it selects from what is already there (mutation creates the new variation).

3. **evolution**

   Selection and genetic drift change allele frequencies — that is evolution at the genetic level.

4. **humans choose which organisms breed, rather than nature**

   In artificial selection, people choose the breeders to pass on useful features; in natural selection, the environment selects.

5. **the gene pool of a population changing over generations, forming new species**

   Evolution is the change in a population's gene pool over time, which can lead to new species.

6. **a smooth range controlled by many genes plus the environment**

   Continuous variation (e.g. height) is a smooth range from many genes; discontinuous is separate groups from few genes.

7. **those best adapted, which survive and reproduce**

   The best-adapted individuals survive, reproduce, and pass on their helpful alleles, which spread over generations.

8. **artificial**

   Artificial selection (selective breeding) is the same mechanism as natural selection — but humans, not the environment, do the selecting.

9. **True**

   It can be allopatric (geographical barrier) or sympatric (same area).

10. **True**

    A narrow gene pool concentrates harmful recessive alleles and lowers the population's resilience.

A-LEVEL BIOLOGY • EXERCISE SHEET

17.1 Variation

Name: _____ Class: _____ Date: _____ **Total: 22 marks**

KEY WORDS variation 变异 • genetic 遗传 • continuous 连续 • discontinuous 不连续 • continuous variation 连续变异

Objective

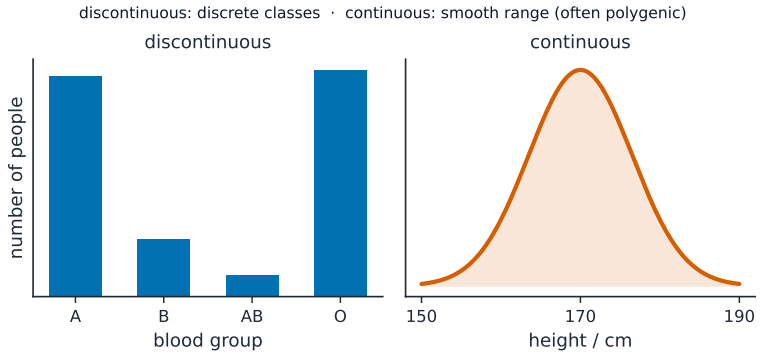
Build the skills to answer exam questions on **variation** — **genetic** 遗传 and **environmental** 环境 causes, **continuous** 连续 and **discontinuous** 不连续 variation, and comparing samples with the **t-test**.

You must be able to:

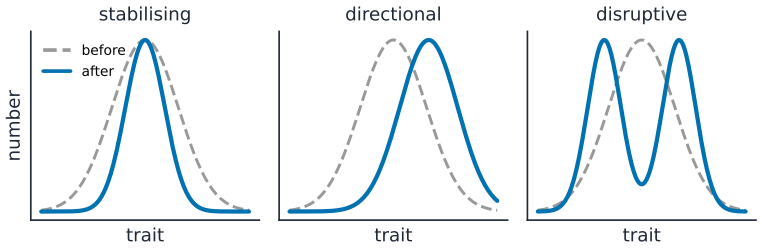
- explain genetic, environmental and combined causes of phenotypic variation
- explain continuous and discontinuous variation and their genetic basis
- use a t-test to compare the means of two samples

1 Worked examples

Causes and patterns of variation



Discontinuous variation falls into separate groups; continuous variation is a smooth range



directional: favors one extreme · stabilising: favors mean · disruptive: favors both extremes

Variation is acted on by the types of selection

- **genetic only** — set by alleles (e.g. blood group); **environmental only** — set by surroundings (e.g. a scar); most features are a **combination** (e.g. height).
- **discontinuous**: separate categories, controlled by one or a few **genes** 基因, little environmental effect.
- **continuous**: a smooth range, controlled by **many genes** plus the environment.

The t-test

The **t-test** compares the **means** of two samples to see whether their difference is real or just chance. A large t (beyond the critical value) means the difference is significant; a small t means it could be due to chance.

2 Practice

2.1 State whether each is continuous or discontinuous variation: human height; ABO blood group. [2]

2.2 Give **one** example of a feature caused by both genes and the environment. [1]

3 Exam-style questions

3.1 Which feature shows discontinuous variation? [1]

- **A** body mass
- **B** ABO blood group
- **C** height
- **D** skin colour

3.2 Continuous variation is usually caused by: [1]

- **A** a single gene only
- **B** many genes together, plus the environment
- **C** the environment only
- **D** a chromosome mutation

3.3 A student measures the heights of plants grown in sun and in shade and wants to know if the difference in mean height is significant.

(a) Name a statistical test the student should use. [1]

(b) The calculated t value is greater than the critical value at $p = 0.05$. State what this tells the student. [2]

3.4 Explain why two genetically identical plants can still differ in height. [2]

3.5 A student measured the height of 100 plants of one species growing in a field, and the number of plants of each of four flower colours.

(a) State, with a reason, whether height and flower colour show continuous or discontinuous variation. [4]

(b) Describe the causes of the variation in each characteristic. [3]

(c) The heights gave a mean of 42.0 cm and a standard deviation of 6.0 cm. Explain what the standard deviation tells you, and state approximately what percentage of the plants lie between 36.0 cm and 48.0 cm. [3]

(d) Explain why a t-test, rather than a comparison of means alone, would be used to compare this field with a second one. [2]

Solutions

2.1 height — continuous; ABO blood group — discontinuous.

2.2 e.g. height / body mass / skin colour.

3.1 B. ABO blood group falls into separate categories.

3.2 B. Continuous variation is polygenic and affected by the environment.

3.3 (a) the t-test.
(b) the difference between the means is significant; it is unlikely (less than 5% probability) to be due to chance.

3.4 the environment differs (e.g. light, water, minerals); environmental factors affect the phenotype as well as the genes.

3.5 (a) Height is **continuous** — any value within a range is possible, and the data form a smooth distribution with no distinct categories. Flower colour is **discontinuous** — each plant falls into one of a few distinct categories with nothing in between.
(b) Height is **polygenic**, controlled by many genes each of small effect, and it is strongly affected by the environment — light, water and minerals. Flower colour is controlled by one gene, or very few, with little or no environmental influence.
(c) The standard deviation measures the **spread** of the values about the mean — a small value means the plants are closely grouped. 36.0 to 48.0 cm is one standard deviation either side of the mean; for a normal distribution that contains about **68%** of the plants.
(d) The two samples will almost always have slightly different means by chance alone; the *t*-test uses the spread and the sample size to say whether the difference is larger than chance would explain, i.e. whether it is statistically significant.

2 *Exserohilum turcicum* is a fungal pathogen. The growth of the mycelium of the fungus damages the leaves of maize plants, *Zea mays*. Leaf damage reduces crop yield.

(a) (i) Complete Table 2.1 to show **one** structural difference and **one** functional difference between *E. turcicum* and *Z. mays*.

Table 2.1

	E. turcicum	***Z. mays***
structural difference		
functional difference		

[2]

(ii) Describe the principles by which organisms such as *E. turcicum* and *Z. mays* are classified in the taxonomic hierarchy.

.....

.....

.....

.....

.....

.....

.....

..... [4]

(b) Two inbred varieties of maize, SKV50 and CML153, were crossed. The resulting F1 hybrids were self-crossed to produce F2 offspring. The F2 plants were grown, and the percentage area of leaf damage caused by *E. turcicum* was measured.

Fig. 2.1 shows the results for the F2 generation. The arrows show the mean percentage area of leaf damage for the two parent varieties.

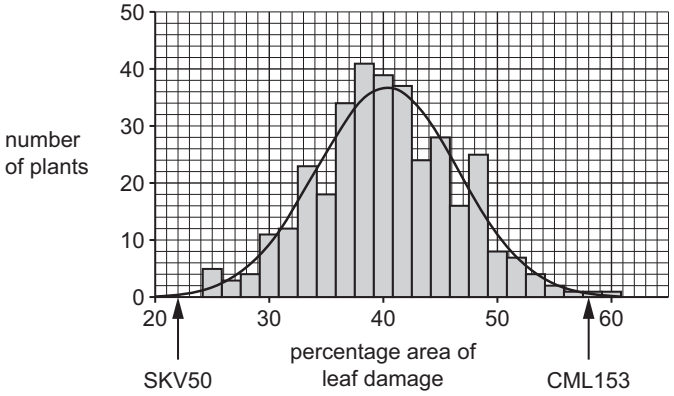


Fig. 2.1

(i) Explain how Fig. 2.1 can be used to determine which parent maize variety shows the greatest resistance to infection by *E. turcicum*.

.....

.....

.....

..... [2]

(ii) State the name of the type of variation shown by the maize F2 generation in Fig. 2.1.

..... [1]

(iii) Explain the **genetic** basis of the type of variation shown by the maize F2 generation.

.....

.....

.....

.....

.....

.....

.....

..... [4]

A-LEVEL BIOLOGY • EXERCISE SHEET

17.2 Natural and artificial selection

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

KEY WORDS natural selection 自然选择 • selective breeding 选择育种 • hardy–weinberg principle 哈迪 • founder effect 奠基者效应 • genetic drift 遗传漂变

Objective

Build the skills to answer exam questions on **natural and artificial selection** — **natural selection** 自然选择, types of selection, the **founder effect** 奠基者效应 and **genetic drift** 遗传漂变, the **Hardy–Weinberg principle** 哈迪-温伯格原理, and **selective breeding** 选择育种.

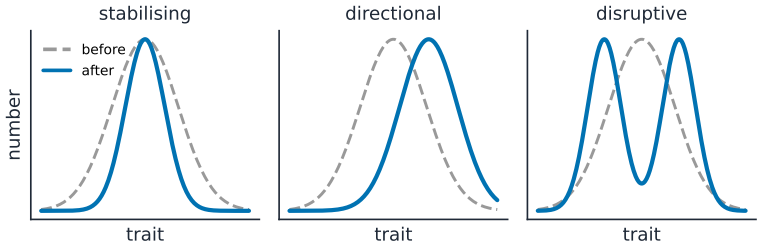
You must be able to:

- explain natural selection and how it acts as a stabilising, directional or disruptive force
- explain the founder effect, genetic drift and the bottleneck effect
- use the Hardy–Weinberg principle and describe selective breeding

1 Worked examples

Natural selection and its forces

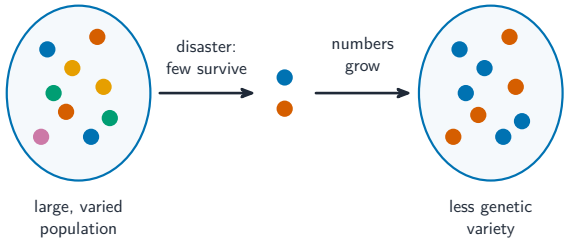
Populations make more offspring than can survive, so they **compete** 竞争; the best **adapted** 适应 survive, reproduce and pass on their alleles, so helpful alleles become more common.



directional: favors one extreme · stabilising: favors mean · disruptive: favors both extremes

Stabilising favours the average; directional favours one extreme; disruptive favours both extremes

Changing allele frequencies



A bottleneck (or founder effect) leaves few alleles; genetic drift changes frequencies by chance

Antibiotic resistance is natural selection: a chance mutation makes some bacteria resistant; the antibiotic kills the rest; the resistant survivors reproduce.

Hardy–Weinberg

With $p + q = 1$ and $p^2 + 2pq + q^2 = 1$, you can find allele and genotype frequencies (when the population is large, mating random, and there is no mutation, migration or selection).

Selective breeding

Humans choose which organisms breed, so useful features (e.g. disease resistance, milk yield) increase

2 Practice

2.1 Name the type of selection that favours the average and removes the extremes. [1]

2.2 State **two** conditions under which the Hardy–Weinberg principle holds. [2]

3 Exam-style questions

3.1 A change that favours one extreme of a range, shifting the mean, is: [1]

- **A** stabilising selection
- **B** directional selection
- **C** disruptive selection
- **D** genetic drift

3.2 The bottleneck effect reduces a population's: [1]

- **A** mutation rate
- **B** genetic variety (allele variety)
- **C** birth rate
- **D** number of chromosomes

3.3 In a population, the recessive phenotype (genotype q^2) occurs in 16% of individuals.

(a) Calculate the frequency of the recessive allele, q. [2]

(b) Calculate the frequency of heterozygotes ($2pq$). [2]

3.4 Explain how the overuse of antibiotics leads to a resistant bacterial population, using the idea of natural selection. [3]

3.5 A population of moths on an island is normally pale. After a volcanic eruption darkened the tree bark, the proportion of dark moths rose from 3% to 78% in twelve generations.

(a) Explain this change in terms of natural selection, in the correct order. [5]

(b) Name the type of selection shown, and describe how it differs from stabilising selection. [3]

(c) A small group of these moths is blown to a second, unaffected island and founds a new population there. Name and explain the effect this is likely to have on that population's allele frequencies. [3]

(d) Explain **one** way in which artificial selection differs from natural selection. [2]

Solutions

2.1 stabilising selection.

2.2 any two: large population; random mating; no mutation; no migration; no natural selection.

3.1 B. Directional selection shifts the mean towards one extreme.

3.2 B. A bottleneck leaves few survivors, reducing allele variety.

3.3 (a) $q^2 = 0.16$, so $q = \sqrt{0.16} = 0.4$.
(b) $p = 1 - q = 0.6$; $2pq = 2(0.6)(0.4) = 0.48$.

3.4 a chance mutation makes some bacteria resistant; using the antibiotic kills the non-resistant bacteria; the resistant ones survive and reproduce, so the resistant allele becomes more common.

3.5 (a) The dark form already existed in the population, produced originally by **mutation**, so there was pre-existing variation; after the eruption the pale moths were conspicuous on dark bark and the dark moths were camouflaged; birds ate proportionally more pale moths — this is the **selection pressure**; the surviving dark moths were more likely to reach reproductive age and pass on the allele for dark colour; so the frequency of that allele rose generation by generation.
(b) **Directional** selection. It favours one extreme of the range, so the mean shifts; stabilising selection favours the intermediate and acts against both extremes, so the mean stays put and the variation narrows.
(c) The **founder effect**. The few individuals that arrive carry only a small, unrepresentative sample of the original population's alleles; so allele frequencies in the new population differ from the old one, some alleles are missing altogether, and genetic diversity is low.
(d) In artificial selection a **human** chooses which individuals breed, according to a trait people want; in natural selection the environment does the selecting, and the trait that spreads is whichever improves survival and reproduction.

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.

.....

.....

.....

.....

.....

.....

.....

..... [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.

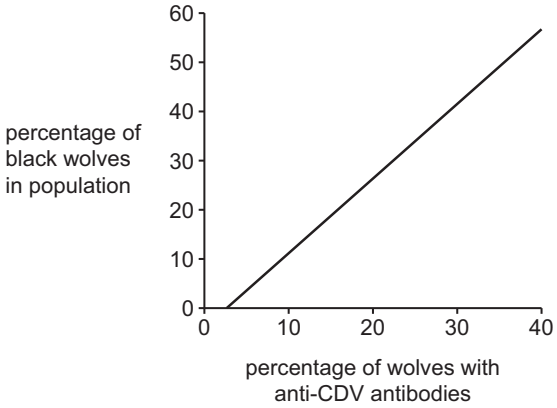


Fig. 4.1

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.

.....

.....

.....

.....

.....

.....

..... [4]

[Total: 11]

4 The evolution of antibiotic resistance in bacteria has occurred as a result of natural selection.

(a) Name **two** ways in which a bacterium can become resistant to an antibiotic.

.....

..... [2]

(b) The World Health Organisation regularly analyses bacterial DNA sequence data.

Suggest **one** way in which this contributes to solving the problem of antibiotic resistance in bacteria.

.....

.....

..... [1]

(c) Some infectious bacterial diseases are treated with the antibiotic streptomycin.

If a person does **not** finish the prescribed course of streptomycin, bacteria are more likely to become resistant to the antibiotic.

Explain why a streptomycin-resistant strain of bacteria is more likely to develop as a result of natural selection when a person does **not** complete the prescribed course of antibiotics.

.....

.....

.....

.....

.....

..... [3]

[Total: 6]

Question 5 starts on page 12.

5 The Galápagos is a group of islands with a high biodiversity.

(a) *Geospiza fortis* is one of the many species of finch that live on the Galápagos islands. On one of the Galápagos islands, Daphne Major, scientists measured the bill size (length, width and depth) of individuals in a population of *G. fortis* over several years.

Fig. 5.1 shows a *G. fortis* female.

Fig. 5.1

G. fortis feed on seeds. Seed size and bill size vary. It is easier for *G. fortis* with smaller bills to eat small seeds and for *G. fortis* with larger bills to eat large seeds.

In 1977, a drought occurred on Daphne Major, which resulted in a large decrease in the availability of seeds, particularly small seeds.

Scientists observed an increase in the mean bill size in the *G. fortis* population on Daphne Major after the drought.

(i) State the type of natural selection that occurred in the *G. fortis* population on Daphne Major as a result of the drought.

..... [1]

(ii) Fig. 5.2 shows the distribution of bill size in the *G. fortis* population before the drought in 1977.

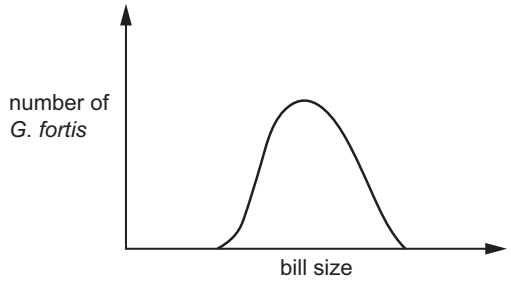


Fig. 5.2

Sketch a new curve on Fig. 5.2 to snow the distribution of bill size in the *G. fortis* population after the drought.

[1]

(b) *G. fortis* also lives on Santa Cruz, another island in the Galápagos.

Most of the seeds available for *G. fortis* to eat on Santa Cruz are either small or large. There are few intermediate sizes of seed.

Fig. 5.3 shows the distribution of bill size in the *G. fortis* population on Santa Cruz.

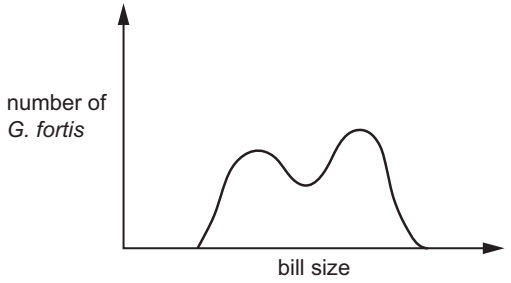


Fig. 5.3

Explain how natural selection in the *G. fortis* population on Santa Cruz has resulted in the distribution in bill size shown in Fig. 5.3.

.....

.....

.....

.....

.....

.....

.....

.....

.....

..... [4]

(c) Scientists studied the effect of an increase in the number of tourists each year on the number of invasive alien species present in the Galápagos islands.

The scientists recorded the number of invasive alien species present each year in the Galápagos islands over many years. The scientists also recorded the number of tourists visiting the islands each year.

Fig. 5.4 shows a graph of the data for five different years during the study.

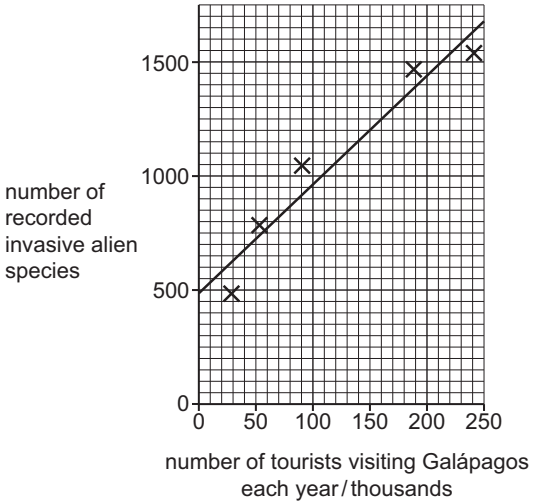


Fig. 5.4

(i) The scientists carried out a Pearson's linear correlation calculation for the data in Fig. 5.4.

The scientists calculated an ***r*** value of 0.930.

The scientists concluded that there is a significant correlation between the number of tourists visiting the Galápagos islands each year and the number of invasive alien species present.

Table 5.1 shows a probability table of critical values for Pearson's linear correlation.

Table 5.1

number of observations	probability level (*p*)	
	0.05	**0.01**
3	0.997	1.000
4	0.950	0.990
5	0.878	0.959
6	0.811	0.917
7	0.754	0.875

With reference to Table 5.1, explain why the ***r*** value of 0.930 indicates that there is a significant correlation between the number of tourists and the number of invasive alien species.

.....

.....

.....

..... [2]

(ii) Invasive alien species are thought to have contributed to extinctions recorded in the Galápagos islands.

Discuss the negative effects of introducing invasive alien species to an ecosystem.

.....

.....

.....

.....

.....

.....

.....

.....

..... [4]

(iii) State **two** factors, other than competition and the introduction of invasive alien species, that can cause extinction.

1 .....

2 .....

[2]

[Total: 14]

A-LEVEL BIOLOGY • EXERCISE SHEET

17.3 Evolution

Name: _____ Class: _____ Date: _____ **Total: 24 marks**

KEY WORDS species 物种 • evolution 进化 • sequence 序列 • speciation 物种形成 • allopatric 异域

Objective

Build the skills to answer exam questions on **evolution** — the theory of evolution, **DNA sequence** 序列 evidence, and **speciation** 物种形成 by genetic isolation.

You must be able to:

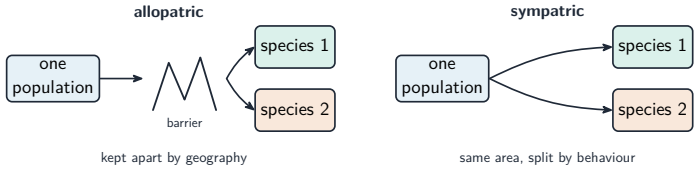
- outline evolution as the change in gene pools producing new species over time
- discuss how DNA sequence data shows evolutionary relationships
- explain allopatric and sympatric speciation

1 Worked examples

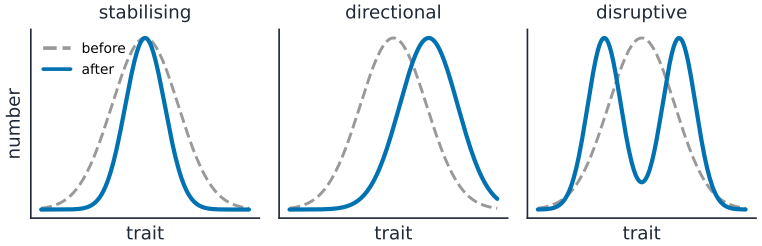
Evolution and DNA evidence

Evolution 进化 is the formation of new **species** 物种 from earlier ones as the **gene pool** 基因库 (all the alleles in a population) changes over generations. **DNA sequence data** shows relationships: the more similar two species' DNA, the more recently they shared a common ancestor.

Speciation



Allopatric speciation needs a physical barrier; sympatric speciation happens in the same area



directional: favors one extreme · stabilising: favors mean · disruptive: favors both extremes

Selection drives evolution and speciation

Speciation happens when two populations become genetically **isolated** 隔离 so they can no longer interbreed:

- **allopatric** 异域 — separated by a **geographical** barrier (sea, mountain).
- **sympatric** 同域 — in the same area, separated by behaviour or way of life.

2 Practice

2.1 Define **evolution** in terms of the gene pool. [2]

2.2 State the difference between allopatric and sympatric speciation. [2]

3 Exam-style questions

3.1 Two species share a very similar DNA base sequence. This suggests they: [1]

- **A** are not related
- **B** shared a recent common ancestor
- **C** live in the same habitat
- **D** have the same number of chromosomes

3.2 A mountain range divides a population into two, which slowly become different species. This is: [1]

- **A** allopatric speciation
- **B** sympatric speciation
- **C** genetic drift
- **D** artificial selection

3.3 Explain how allopatric speciation can lead to the formation of two new species. [4]

3.4 Explain how comparing DNA sequences can be used to show how closely related different species are. [2]

3.5 Two populations of a fish species live in one lake. One feeds at the surface, the other on the bottom. They breed at different depths and no longer interbreed, although in the laboratory they can produce fertile young.

(a) Explain, using the biological definition of a species, whether these are one species or two. [3]

(b) Name the type of speciation occurring here, and explain how it differs from the other type. [3]

(c) Describe how DNA sequence data could be used to estimate how long ago the two populations began to diverge. [3]

(d) Explain why comparing DNA sequences is better evidence of relatedness than comparing external appearance. [3]

Solutions

2.1 evolution is the change in the gene pool / allele frequencies of a population over many generations, leading to new species.

2.2 allopatric — populations separated by a geographical barrier; sympatric — populations in the same area separated by behaviour/ecology.

3.1 B. Very similar DNA suggests a recent common ancestor.

3.2 A. A geographical barrier causing speciation is allopatric.

3.3 a geographical barrier separates the population into two; each population faces different conditions and undergoes different selection / mutations / drift; their gene pools change in different ways; eventually they are so different they can no longer interbreed to produce fertile offspring — two species.

3.4 the more similar the DNA base sequences, the more recently the species shared a common ancestor; large differences indicate they diverged longer ago.

3.5 (a) A species is a group of organisms with similar features that can **interbreed to produce fertile offspring**. These populations can do so in the laboratory, so by that definition they remain one species; but they are reproductively isolated in the wild, so they are diverging and would be described as incipient species or subspecies.
(b) **Sympatric** speciation — the two populations occupy the same geographical area; they are separated by a behavioural or ecological barrier, here breeding depth. Allopatric speciation instead begins with a **physical** barrier such as a river or mountain that geographically separates the populations.
(c) Sequence the same gene in both populations and count the number of base differences; mutations accumulate at a roughly constant rate over time — the molecular clock; so the number of differences, divided by that rate, estimates the time since the populations shared a common ancestor.
(d) Appearance can mislead in two opposite ways: unrelated species may evolve similar features because they face the same selection pressure — convergent evolution; and closely related species may look very different if their environments differ. DNA is the actual heritable material, so the number of sequence differences reflects ancestry directly, and it can compare organisms with no visible features in common at all.

4 Alleles are alternative forms of a gene. For example, there may be a dominant allele, **T**, and a recessive allele, **t**, at the same gene locus.

The relative frequency of each allele of a gene in a population can change over time due to factors such as selection, genetic drift and the bottleneck effect.

(a) Fig. 4.1 shows the relative frequency of the **T** allele in a population of 50 individuals over 20 generations.

At generation 0, the number of **T** alleles and the number of **t** alleles in the population was equal, so the relative frequency of each allele was 0.5. The relative frequencies of the two alleles of the gene add up to 1.

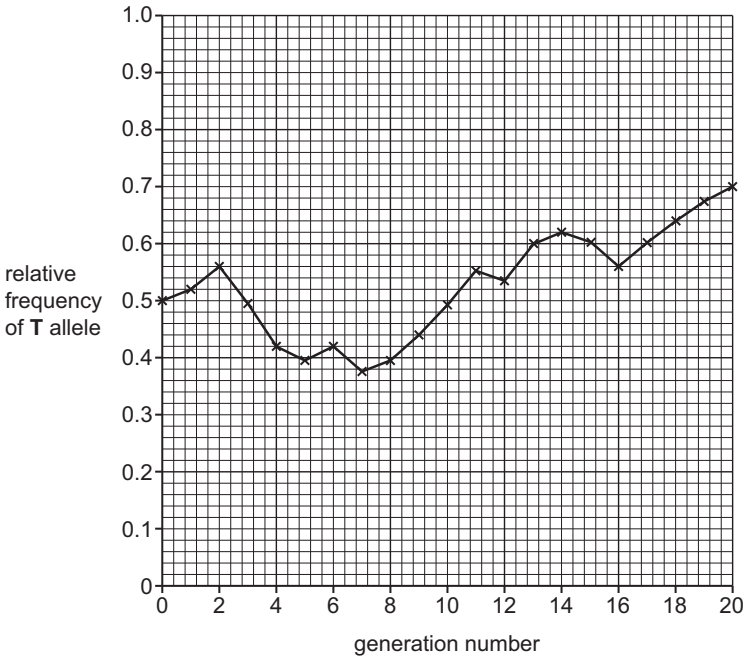


Fig. 4.1

(i) With reference to Fig. 4.1, state the relative frequency of the **t** allele after 20 generations.

..... [1]

(ii) Suggest possible explanations for the change in the relative frequency of the **T** allele between generation 0 and generation 13.

.....

.....

.....

.....

.....

.....

.....

.....

.....

..... [4]

(b) The relative frequency of the **T** allele in a population of only 10 individuals was determined over 20 generations. The environmental conditions remained the same throughout the experiment.

Fig. 4.2 shows the results.

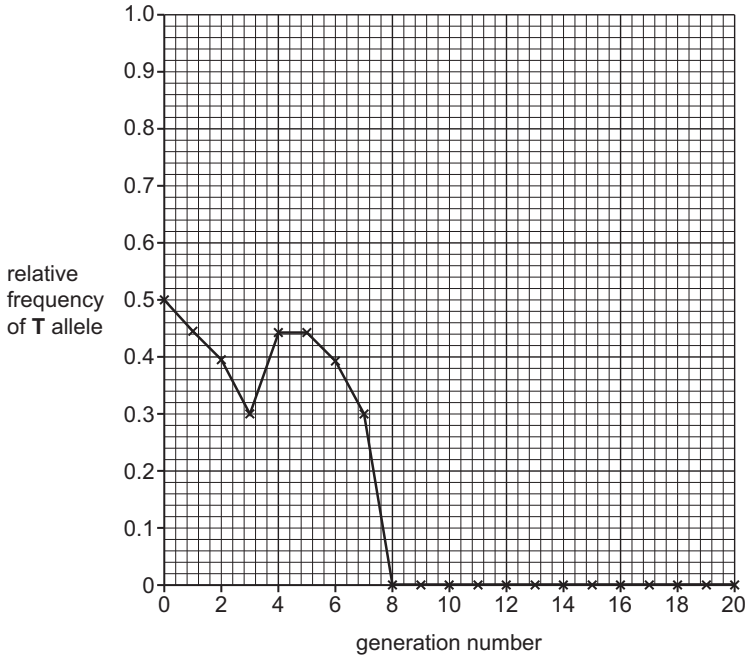


Fig. 4.2

Explain why Fig. 4.2 shows a different result from Fig. 4.1.

.....

.....

.....

.....

.....

.....

.....

..... [3]

(c) State the name of the principle that can be used to calculate relative frequencies of two alleles by counting the numbers of organisms in the population showing dominant and recessive phenotypes.

..... [1]

[Total: 9]

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.

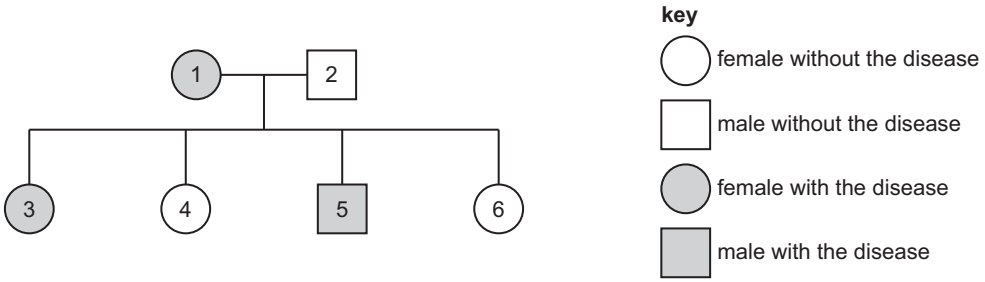


Fig. 3.1

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 .....

.....

.....

.....

.....

[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.

.....

.....

.....

.....

.....

.....

..... [3]

(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.

.....

.....

.....

.....

.....

.....

.....

..... [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.

.....

.....

..... [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.

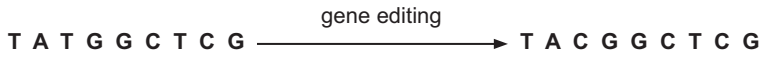


Fig. 3.2

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.

.....

.....

.....

.....

....

..... [3]