DP IB Biology: HL

Your notes

10.3 Gene Pools & Speciation

Contents

Your notes

10.3.1 Gene Pools

Gene Pools

- A gene pool consists of all the genes and their different alleles, present in an interbreeding population
- Some populations of the same species are geographically isolated from each other
  - So **multiple gene pools** can exist for a species
- Individuals in a population tend to have **common characteristics** and resemble each other
  - Two geographically-isolated population may have **different characteristics** whilst still being the same species
  - This is one basis for **speciation**
- A consideration of all the genes (and alleles thereof) in a population is important as that will govern the **genomes of the next generation**
  - This collection of genes and alleles is commonly called the gene pool

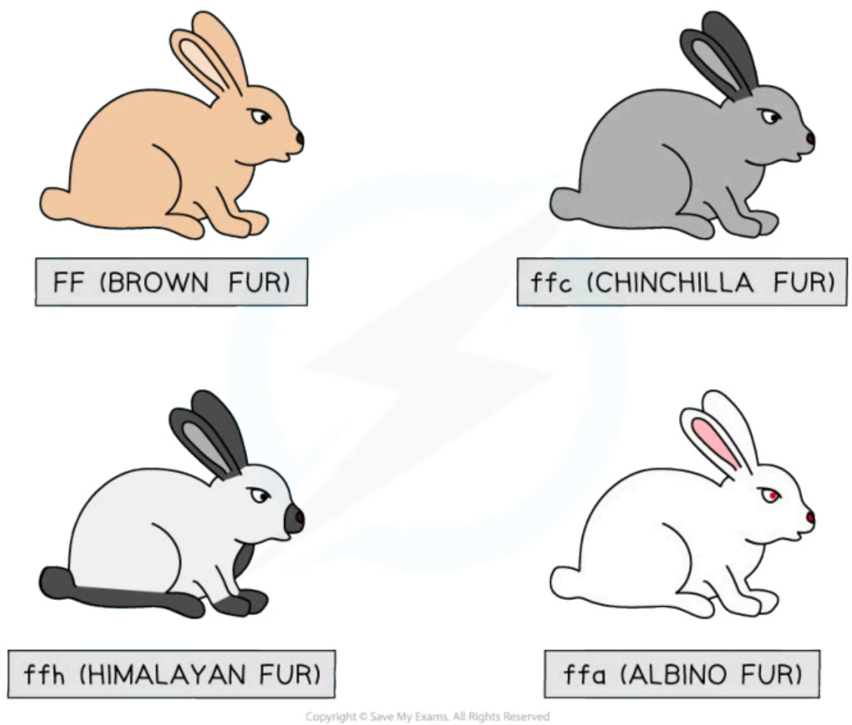
Calculating allele frequencies

- Allele frequencies are defined as the **relative abundance** of alleles for a particular gene
- Allele frequency is calculated by dividing the number of times the **allele of interest** is observed in a population by the total number of copies of **all the alleles** at that particular genetic locus in the population
  - Allele frequencies are expressed as a **number between 0.0 and 1.0** or as percentages **0 – 100%**
    - We can think of this as a **probability** that an allele chosen at random in a given gene will be a particular allele of interest
  - The frequencies of all the alleles for a particular gene **must add up to 1**

Your notes

Worked example

In rabbits, wild-type animals have brown fur, governed by a dominant allele **F**. However, 3 recessive alleles exist, each of which gives a different fur colour and coverage.



Wild-type brown rabbits (FF) and three separate recessive-allele-containing rabbits (ffc, ffh and ffa alleles)

Four separate alleles comprise the gene pool that determines fur colour in this population:
F = brown fur
$\mathbf{f_c}$ = chinchilla fur
$\mathbf{f_h}$ = Himalayan fur
$\mathbf{f_a}$ = albino fur

The albino fur allele ($\mathbf{f_a}$) is four times more frequent in this population than the Himalayan fur allele ($\mathbf{f_h}$). Use this information to complete the table below.

Your notes

Phenotype	Allele	Allele frequency
Brown	F	0.84
Chinchilla	f_c	0.06
Himalayan	f_h	
Albino	f_a	

Step 1: Calculate the sum of the allele frequencies given

Allele frequencies ($F + f_c$) = 0.84 + 0.06 = 0.90

Step 2: Work out the sum of the remaining allele frequencies

All possible allele frequencies must add up to 1, so allele frequencies ($f_h + f_a$) = 1.0 - 0.9 = 0.1

Step 3: Apply the 4:1 ratio of albino allele: Himalayan allele given in the question

0.1 split in a 4:1 ratio is 0.08 : 0.02 for the alleles $f_a : f_h$

Step 4: Complete the table with these frequencies

Phenotype	Allele	Allele frequency
Brown	F	0.84
Chinchilla	f_c	0.06
Himalayan	f_h	0.02
Albino	f_a	0.08
		1.00

Stable gene pools

- Populations retain a **stable** gene pool under the following conditions
  - The population is **large**

- Each individual in the population has an **equal chance of mating**
- That matings are **random**
- There are **no selective pressures** acting upon individuals based on their phenotype
- A stable gene pool means that a population is **not evolving**

Your notes

Examiner Tip

Avoid confusing **allele frequency** with **phenotype frequency**; they mean very different things. An allele can have a high frequency but if it is recessive, it may only be expressed in a minority of the phenotypes of the population.

10.3.2 Evolution

Your notes

The Process of Evolution by Natural Selection

- Evolution requires that **allele frequencies change** with time in populations
- Evolution is defined as the **cumulative change in the heritable characteristics (genes)** of a population over time
- Organisms cannot change their species' phenotype significantly without an **underlying change in their genetic makeup**
- The factors that drive evolution are
  - **Mutations** causing new alleles to come into being
  - **Selection pressures** that favour the existence of certain alleles and oppose that of others
- A key consideration is that **evolution has no purpose**
  - There is **no conscious change** of genetic makeup in order to take advantage of changes in conditions
  - Mutations and selection pressures occur entirely at **random**
  - A species only evolves by virtue of a lucky **combination of advantageous alleles and selection pressures**
  - Organisms developing other alleles that put an affected individual at a selective disadvantage **will not survive** to reproduce and pass on those alleles
- Changes in allele frequencies can sometimes be referred to as genetic drift
- Evolution can happen within a species **before** speciation **occurs**
  - An example is the many dog breeds that all exist within the same species, *Canis familiaris*
  - Whilst many breeds have been selectively bred artificially for aesthetic reasons by humans (or to perform valuable tasks like seeing-eye dogs), most common dog breeds are capable of interbreeding to produce fertile offspring, often referred to as mongrels
- **Population size** has an effect on evolution
  - In a small population, random events such as climate change can have a **dramatic effect** on the frequency of alleles
  - By contrast, in a large population, there is **more capacity to absorb small fluctuations** in allele frequency
- When an allele is put under selective pressure, its frequency drops and sometimes falls to zero as a more advantageous allele becomes more abundant, or becomes the only allele in the gene pool

Example of changes in allele frequency driving evolution

- The **peppered moth** (*Biston betularia*) is a well-documented case study of evolution by natural selection
- In the early 18th century in the northern UK, the light grey-winged form of this moth prevailed
  - These moths were **well-camouflaged** against the light-coloured bark and lichens of their host tree species
  - Black-winged moths (which carried the allele for **melanism**) fared badly as they were easy for predators (mainly birds) to spot against the light background

- During the industrial revolution (mid-late 18th Century), soot from coal-burning factories and house chimneys coated many of the trees in the area, **turning them dark or black**
- This led to an **increase in predation of light-winged** (non-melanistic) moths whose camouflage was no longer so effective
- The dark-winged moths became the predominant variety as the **frequency of the melanism allele increased** over successive generations of moths
- Analysis of the populations throughout this period revealed the following changes in allele frequency

Allele	Dominant or Recessive	Allele Frequency / %	
		Before industrialisation (c. 1820 AD)	After industrialisation (c. 1890 AD)
 M (dark, melanism)	Dominant	0.01%	99%
 m (light, non-melanistic)	Recessive	99.99%	1%
		100%	100%

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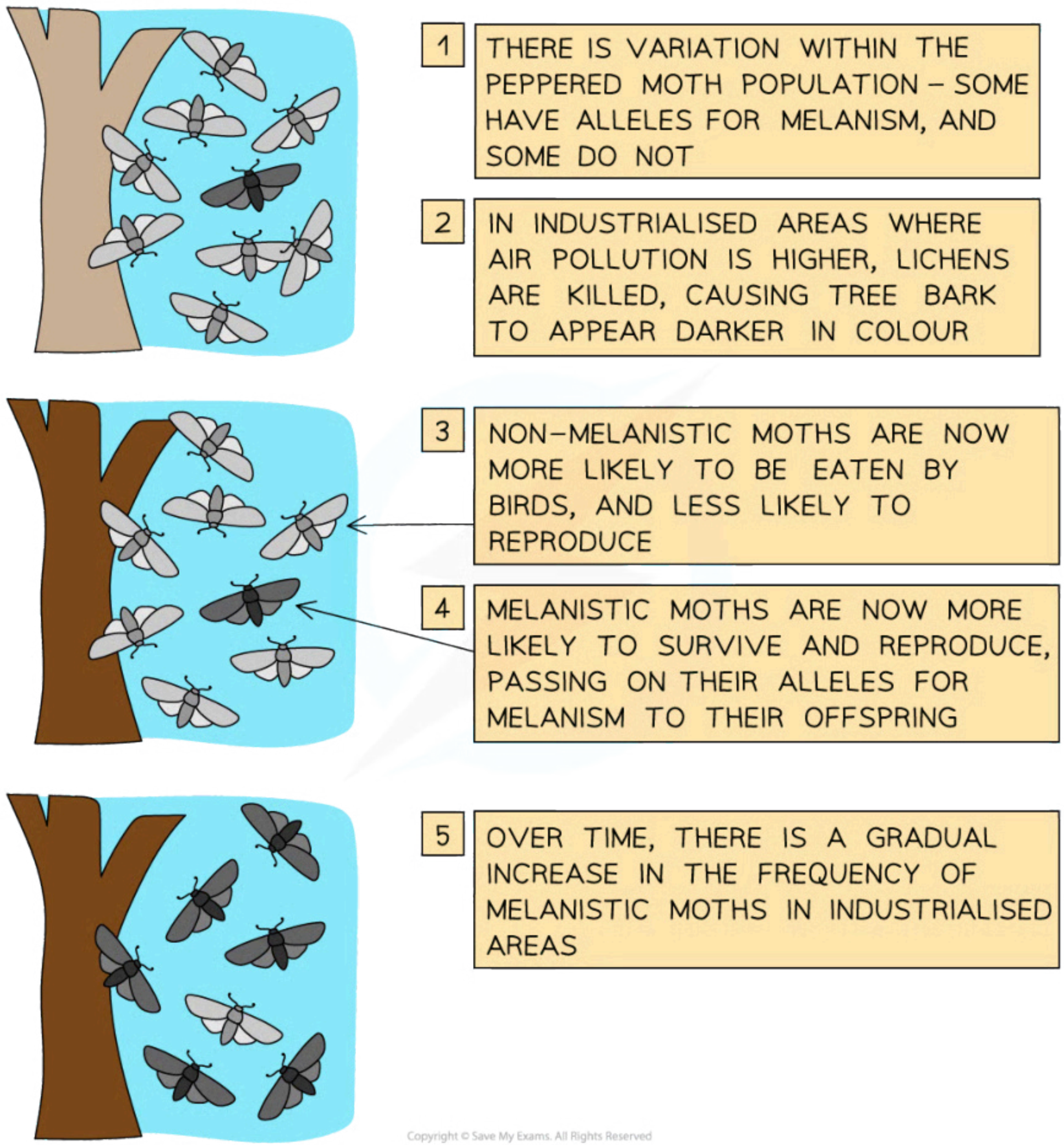

Examiner Tip

Avoid confusing **allele frequency with phenotype frequency**; these are different. In a case where the allele frequency of a recessive trait is 0.4 (40%), this means that the dominant allele must occupy 60% of the gene loci for that gene. The phenotype frequency of the recessive trait will in fact not be 40% because **two copies of the recessive allele need to combine through fertilisation** for the recessive trait to be expressed. The phenotype frequency will therefore be 0.4^2 which is 0.16 (16%). Only 16% of the population will express the recessive trait whereas 40% of the gene loci in the population contain the recessive allele.

Types of Natural Selection

Your notes

- There are three main types of selection:
  - Directional
  - Stabilising
  - Disruptive

Directional selection

- The population changes **towards one extreme** of a range of variation
  - As that extreme becomes better adapted
- This tends to happen when **environmental conditions change**
- For example, a **fall in average temperatures** can affect plants that are not resistant to **frosts** (spells where the air temperature falls below 0°C)
- If there is no allele that can give the species a degree of frost resistance, then the species will **become extinct** in that habitat if cold temperatures endure for more than one generation
- If an allele exists that gives a degree of frost protection, then the species will be able to
  - Survive the frost
  - Go on to reproduce successfully
- Such an allele may code for a new protein that can lower the freezing point of water/cell contents by a few degrees and prevent the formation of damaging ice crystals
- The species has developed a **selective advantage** over other species
- A disadvantageous allele **does not have to be fatal** to an individual organism
- However, it must **prevent the individual from reproducing successfully**
  - Which is effectively the same thing from an evolutionary point of view
  - Because the allele will disappear from the gene pool as the reproductively unsuccessful individuals die
- The species can **change its genome abruptly** by directional selection

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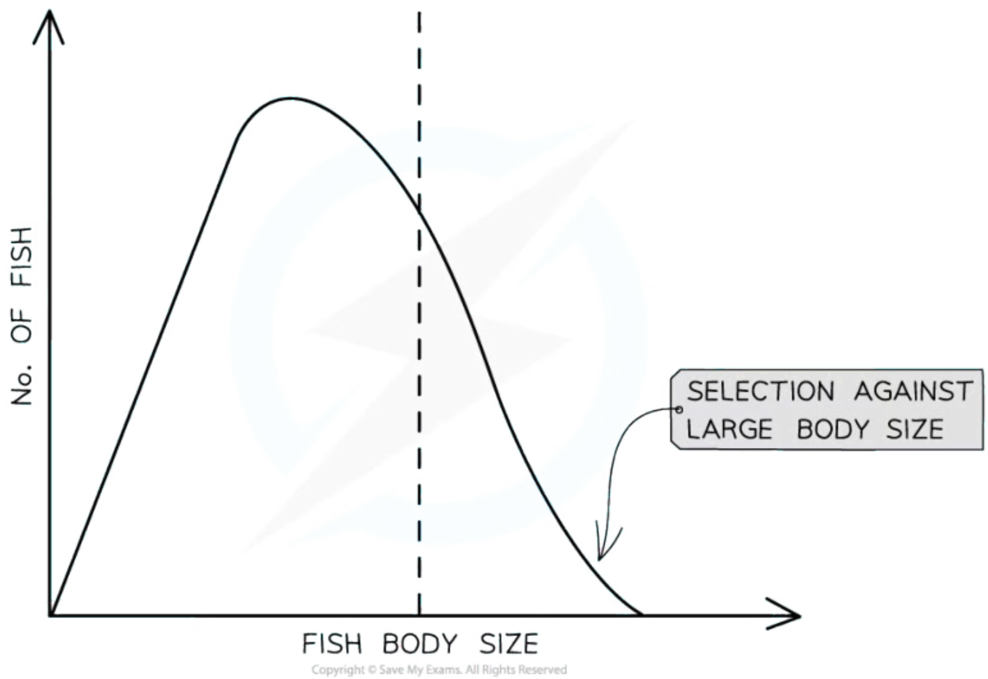
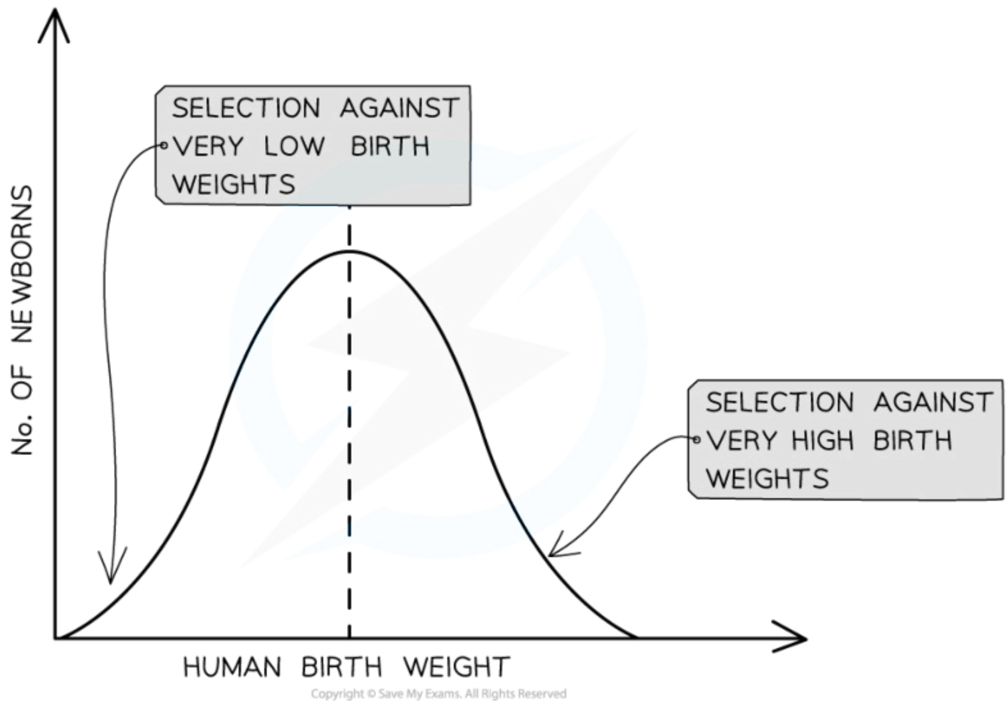


Directional selection acting on fish body size. Increases in ocean temperatures are selecting for smaller body sizes in fish. Warmer seas cause fish metabolism to speed up and so increase their need for oxygen (oxygen levels are lower in warmer seas). Larger fish have greater metabolic needs than smaller fish, and so they feel the effect of increased temperatures more strongly.

Stabilising selection

- Selects **in favour of the average individual** in a population
- Occurs when environmental conditions are stable / do not change
- Selection tends to favour individuals with a range of alleles whose characteristic is the **most advantageous**
- Stabilising selection is the **most common form** of natural selection
- An example is the coat colour of mice
  - The colour stabilises as the one which gives the most camouflage against the surroundings eg. brown fur versus a forest floor
- Birth mass is also an example
  - Where a **normal distribution** clusters around a mean birth mass
  - Too low and too high can lead to problems of survival for an infant
- Stabilising selection
  - Discards **extreme** phenotypes
  - And instead favours the **majority of the population** that is well adapted to their local environment
  - **Decreases diversity** within a population
  - Works mostly on traits that are **polygenic**
  - Is often characterised by a **normal distribution** (a bell-shaped curve)

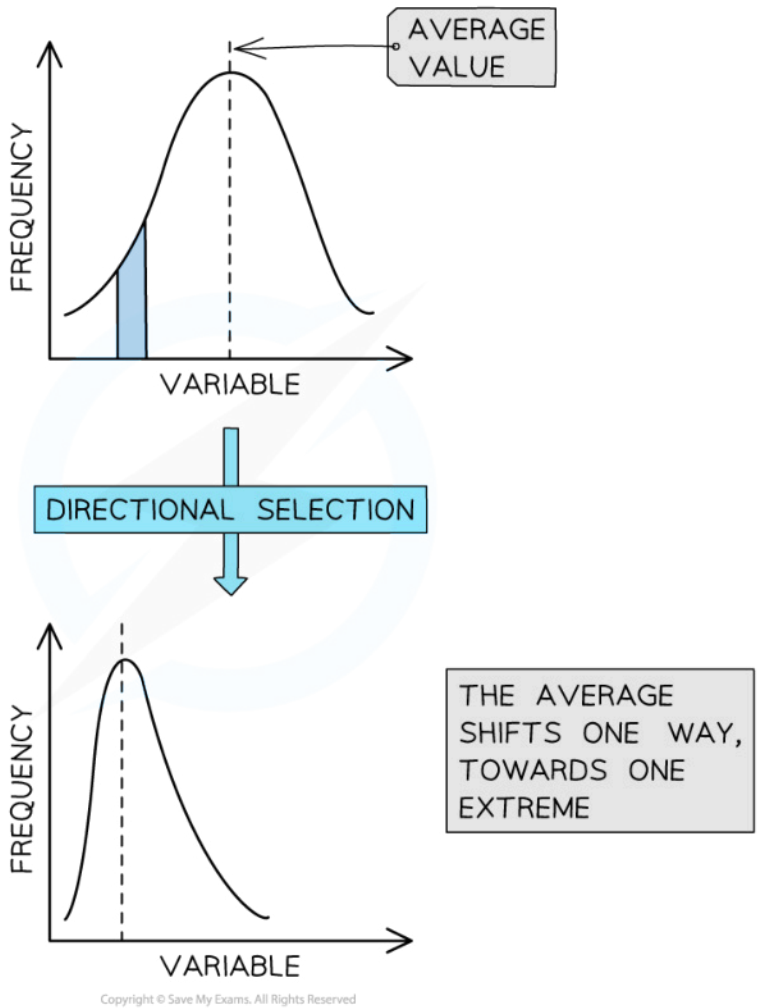
Your notes



Stabilising selection on human birth weight.

Disruptive selection

- Selects **against the average individual** in a population
- Is the rarest of the three forms of natural selection
- Like directional selection, disruptive occurs when **habitats or resources undergo a change**
- Disruptive selection can lead to the **formation of an entirely new species** (speciation)
  - For this reason, is sometimes referred to as '**diversifying** selection'
- **Darwin's finches** in the Galápagos Islands are one of the best-known examples
- Fifteen different species evolved from a common ancestor
- **Multiple types of beaks** have adapted to different food sources over time
- On one island, Santa Cruz
  - Ground finches eat more seeds and some arthropods
  - Tree finches eat more fruits and arthropods
  - Vegetarian finches feed on leaves and fruit
  - Warblers typically eat more arthropods
- When food is **abundant**, their **diets can overlap**
- When food is scarce, these specialisations give each species the ability to compete for a certain type of food better than other species
- This helps each species to occupy its own niche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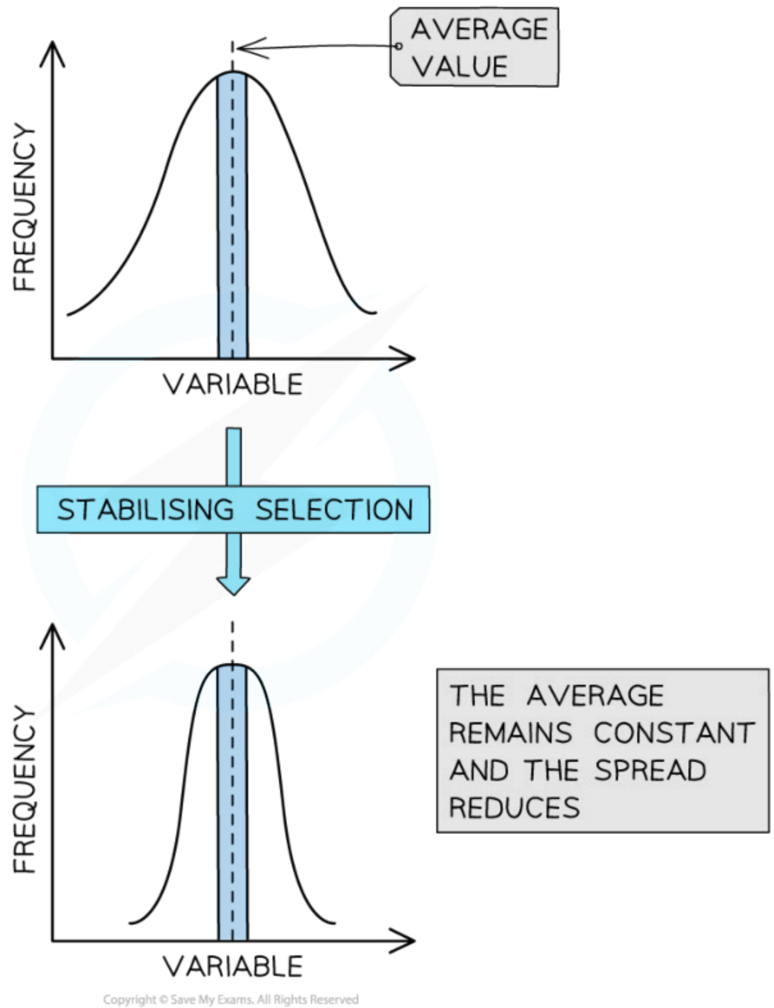
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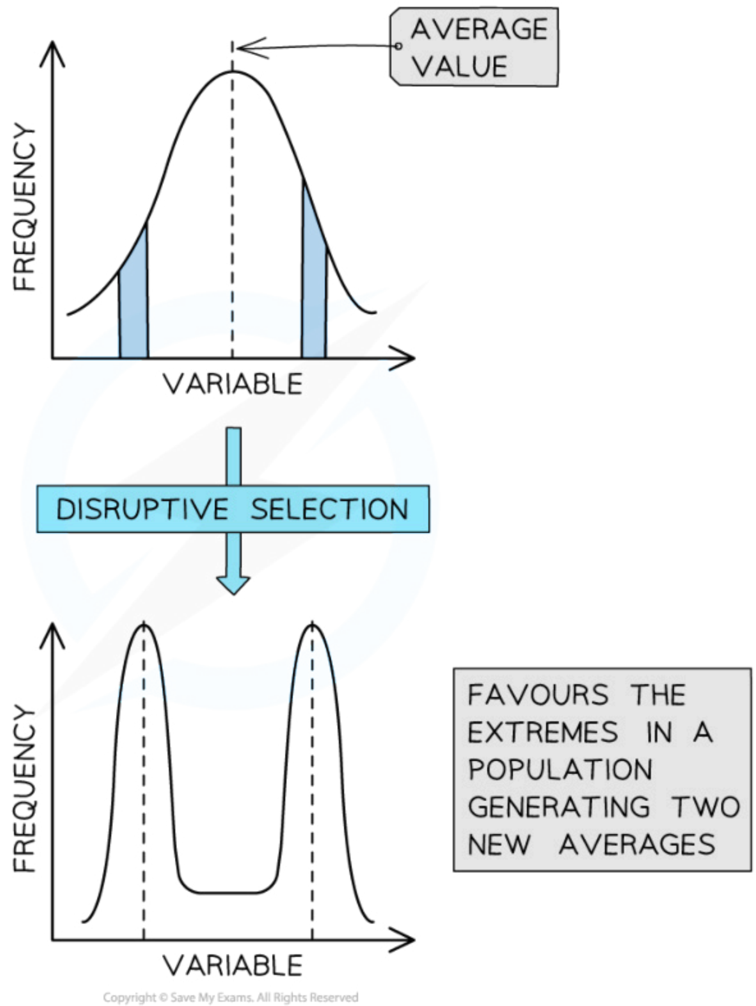
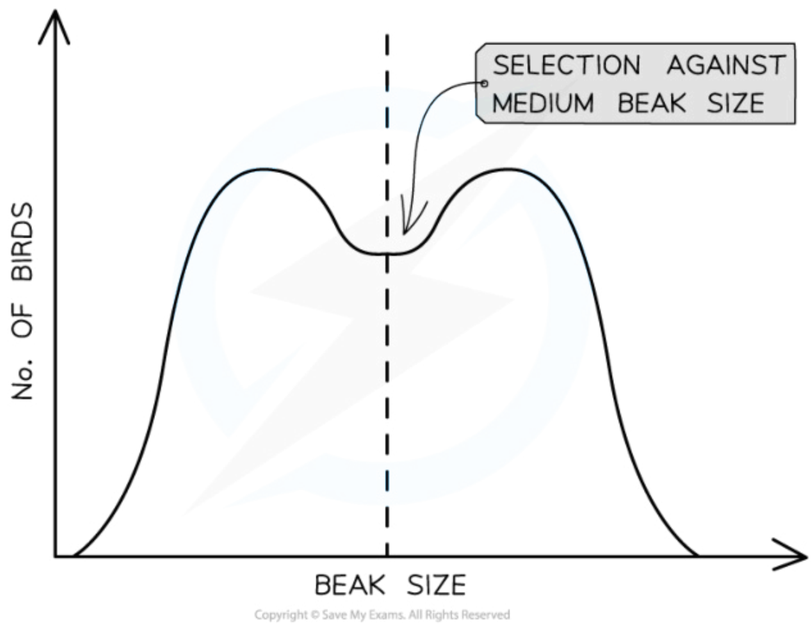


Summary of the 3 main forms of natural selection and their effects on the average phenotype of a population

Your notes



Disruptive selection acting on beak size in a bird population

Examiner Tip

Become familiar with the shapes of the graphs above. They can help you answer questions about the type of selection that is occurring in a population.

Reproductive Isolation

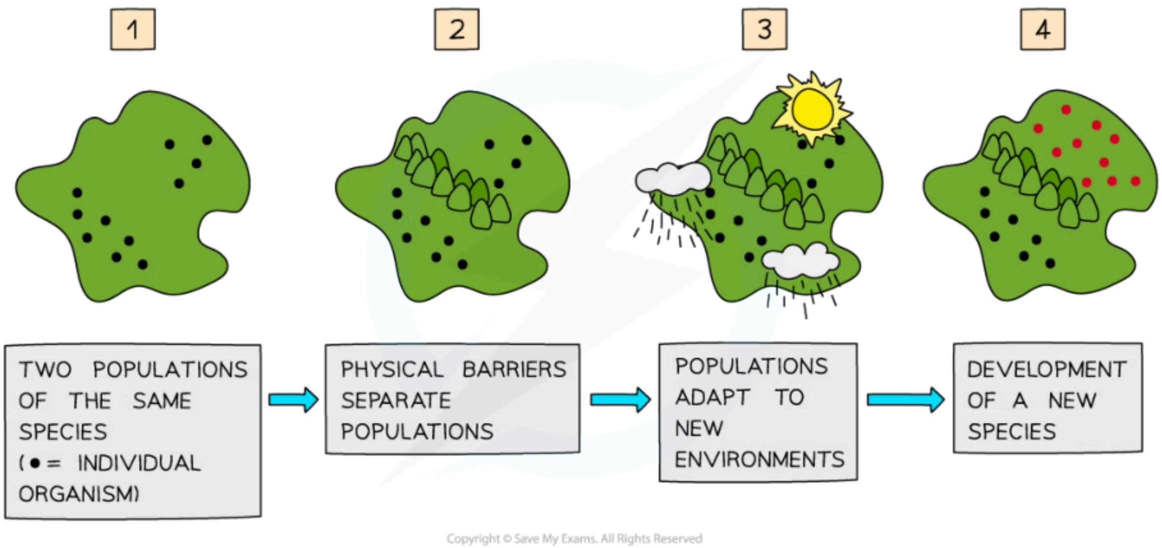
Your notes

- Reproductive isolation of populations can be temporal, behavioural or geographic
- A population of a species can become **isolated from its peers** by various means
    - A **barrier** forms, meaning no interbreeding occurs between the two populations
- For many generations, there may be **little or no variation of the gene pool** within that population
- The nature of the barrier between populations can be temporal, behavioural or geographic
    - Temporal (ie. seasonal)
        - Different groups have **reproductive cycles at different times of the year** so cannot interbreed
        - eg. Changes in **flowering patterns** of plants in different seasons
    - Behavioural
        - Different groups have different rituals or patterns of behaviour meaning that the groups do not recognise each other as 'self'
        - eg. Courtship rituals
    - Geographical
        - eg. When natural or man-made barriers form between two parts of a population, such as a river, freeway, or mountain range
- Over a longer period, the formation of new species (**speciation**) occurs by reproductive isolation ie. separating parts of the population into independently-breeding groups
- Natural selection will **take different paths** according to the differences of biotic and abiotic factors on either side of the divide

Sympatric vs allopatric speciation

- Temporal and behavioural speciation are both examples of **sympatric speciation**, in which separately-developed species can coexist in the **same geographical area** whilst occupying different niches
- Geographic speciation is sometimes called **allopatric speciation**, in which two separate species diverge with **complete spatial separation**

Your notes



Geographical isolation can lead to speciation.

Examiner Tip

A typical exam question will ask you to identify which form of natural selection is taking place from a set of data in the question. Alternatively, you may have to give reasons from the data about why a particular example of natural selection is identified as one of the three forms. Think about how the 'average' individual in a population is affected and that will lead you towards the correct answer.

10.3.3 Speciation

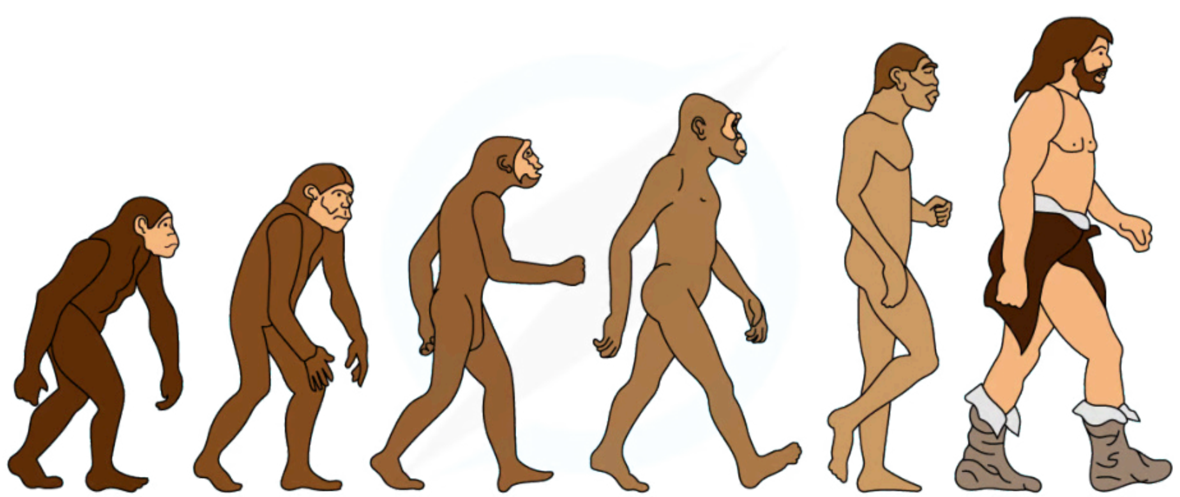
Your notes

Gradualism & Speciation

- Speciation can be defined as the emergence of new and distinct species that are reproductively isolated from other separate species
- Two theories exist to explain the emergence of new species
  - Gradualism
  - Punctuated Equilibrium

Gradualism

- Speciation due to divergence of isolated populations can be **gradual**
- Large changes between species occur due to the **culmination of many small changes** that **accumulate** over time
- Because of the long period of time in which life has existed on Earth (approx 3.5 billion years), one might expect that organisms speciated gradually from their ancestors
- Evidence exists in the fossil record to show that **patterns of evolution can follow the geological cycle**, which consists of long, slow changes that take place over thousands and millions of years
- Charles Darwin originally subscribed to the point of view of gradualism, having observed **vestigial structures** in the fossil record
  - Vestigial structures are observable characteristics that have **no apparent function**
  - They are **residual parts** from a past ancestor that are still inherited but have fallen into disuse
  - Examples of vestigial structures include the **human appendix** and the **wings of flightless birds**

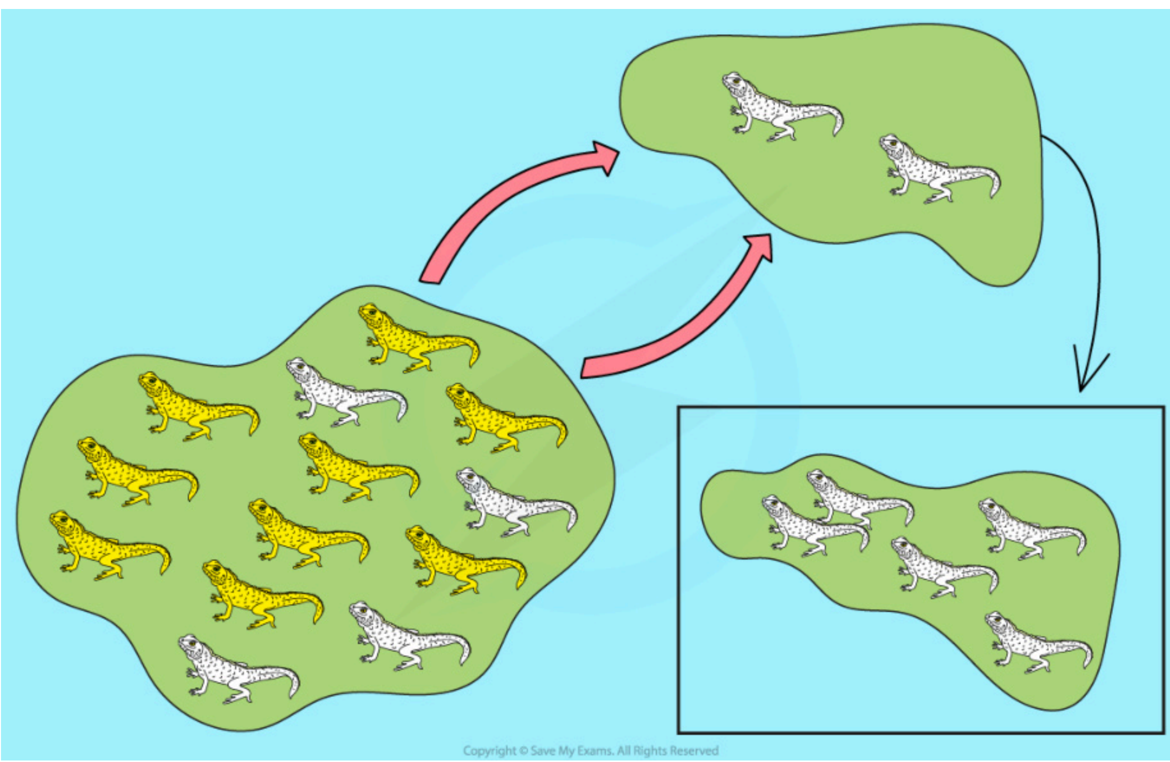


The iconic image of evolution implies gradualism as humans and chimpanzees both evolved from a common ancestor via intermediate species

Your notes

Punctuated Equilibrium

- Speciation can occur **abruptly**
- Punctuated equilibrium implies long periods without appreciable change and short periods of **rapid** evolution
- In the late 19th century, palaeontologists (scientists specialising in the study of life forms that existed in past geological periods) began to notice **anomalies in the fossil record** that **cast doubt** on Darwin and others' theories of gradualism
- One such scientist was William Bateson (who along with Reginald Punnett first observed non-Mendelian inheritance patterns)
- Breaks occurred in the fossil record that revealed **no intermediate species**
- Fossil appeared **relatively unchanged** for long periods of time yet **changed abruptly** at other times
- Sudden **mass extinctions** were observed
  - Cataclysmic events such as huge **volcanic eruptions**, **meteor strikes** and **large-scale gaseous changes to the atmosphere** can cause mass extinctions
  - Some members of the populations that are not adversely affected may **survive** the event
  - These can restart reproduction with a **reduced gene pool**
  - This is called the **Founder Effect**



The Founder Effect as shown by lizards. If the original island was destroyed and only the white (recessive phenotype) lizards move to the new island and so the whole population ends up having the white phenotype.

Examiner Tip

An analogy for **punctuated equilibrium** is a pool-drop river that features long stretches of calm, slow-flowing water punctuated by rapids and waterfalls. **Gradualism** would be represented by a river that flows smoothly down a shallow, uninterrupted gradient out to the ocean.

Your notes

Your notes

10.3.4 Polyploidy & Speciation

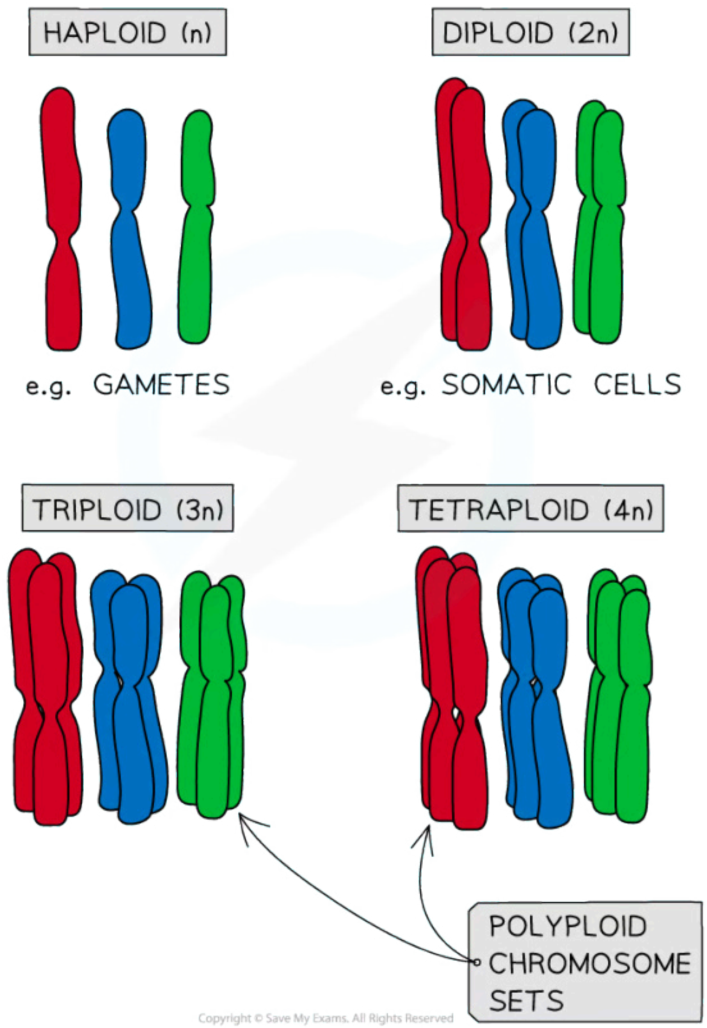
Polyploidy & Speciation

NOS: Looking for patterns, trends and discrepancies—patterns of chromosome number in some genera can be explained by speciation due to polyploidy

- When patterns and trends are observed in nature, scientists seek to find explanations that fit with these observations
- When exceptions to accepted trends are observed in the natural world, it can sometimes mean that established modes of thinking are incorrect, so it is important to consider discrepancies carefully

Polyploidy

- So far, speciation has been explored in the context of mutations and changes to an organism's **existing genome**
  - Where the number of chromosomes remains 2*n* or 2 x the haploid number, but the chromosomes' base sequences alter as new alleles form
- Speciation can also occur through **polyploidy**
- Polyploidy
  - Occurs when an organism has **more than two sets** of homologous chromosomes
  - Is more common in **plants** than in animals
  - Can result from **chromosomal mis-events** eg. abnormalities in mitosis or more commonly, in meiosis
    - For example, the fertilisation of an egg by more than one sperm, or by failure of chromosomes to separate in meiosis I
- A diploid gamete can be formed, which can fertilise with a haploid gamete and produce fertile offspring
- Polyploidy creates a separate taxonomic category within species, often called **breeds or varieties**
- Patterns in chromosome number between organisms (diploid, triploid, tetraploid etc) have been used to explain speciation
- Polyploidy can result in **sympatric speciation** as polyploid and diploid counterparts can coexist in the same geographical area



Chromosome sets showing haploid, diploid and polyploid sets

Uses of polyploidy

Application: Speciation in the genus *Allium* by polyploidy

- Many crop species have been created to be polyploid
  - For example, species in the *Allium* genus
    - Such as onion, garlic, shallots, leeks and chives
    - *Allium porrum* is the cultivated leek and is tetraploid and fertile, so has many advantages over non-polyploid counterparts
- Wild onion (*Allium canadense*) has a diploid number of 2*n* = 14 although polyploid varieties have been generated with 2*n* = 28
- The common onion, (*Allium cepa*) has naturally-occurring polyploid varieties, some of which have been cultivated for agricultural use
- Polyploidy **increases allelic diversity** and **permits novel phenotypes** to be generated

Your notes

  - Having **multiple copies of the same gene** reduces the risk of recessive mutations causing detrimental effects
  - Novel phenotypes can **include improved flavour** and aroma for cooking, a **greater yield** for farmers and **improved pest resistance**
- It also leads to **hybrid vigour** (the tendency of cross-bred individuals to show superior characteristics to those of their parents)

10.3.5 Skills: Comparing Allele Frequencies Between Populations

Your notes

Populations & their Allele Frequencies

Comparison of allele frequencies of geographically isolated populations

- Allele frequency is a term that assigns a **relative frequency** of an allele at a particular gene locus
  - Alleles can vary from each other by as little as one nucleotide
- When a degree of geographic **separation** exists between two populations, this can cause differences in the frequencies of alleles to emerge
- Human allele frequencies vary by **geography** and **ethnicity**
  - Examples of clear-cut allele frequency differences are rare in human populations because of the ease of **travel** and **interbreeding**
  - This leads to a scarcity of **truly isolated** populations
- If there are more than one allele in existence for a particular gene, the respective **allele frequencies must add up to 1**
- Online databases list the frequencies of human alleles
- Alleles are sometimes referred to as **polymorphisms** which just means many (poly-) different forms (-morphisms) of a gene
  - The most common type is called a **single nucleotide polymorphism** (SNP)
- Mathematical formulae such as the **Hardy-Weinberg formula** can be used to calculate phenotype frequencies from allele frequencies and *vice versa*
- Comparing allele frequencies can provide information for
  - Identifying **genetic associations** with particular diseases
  - Estimating the **number of individuals with disease susceptibility** within a population
  - Estimating the level of **drug resistance** in a population
  - Performing **evolutionary and anthropological studies** (eg. tracing the history of humans through time)

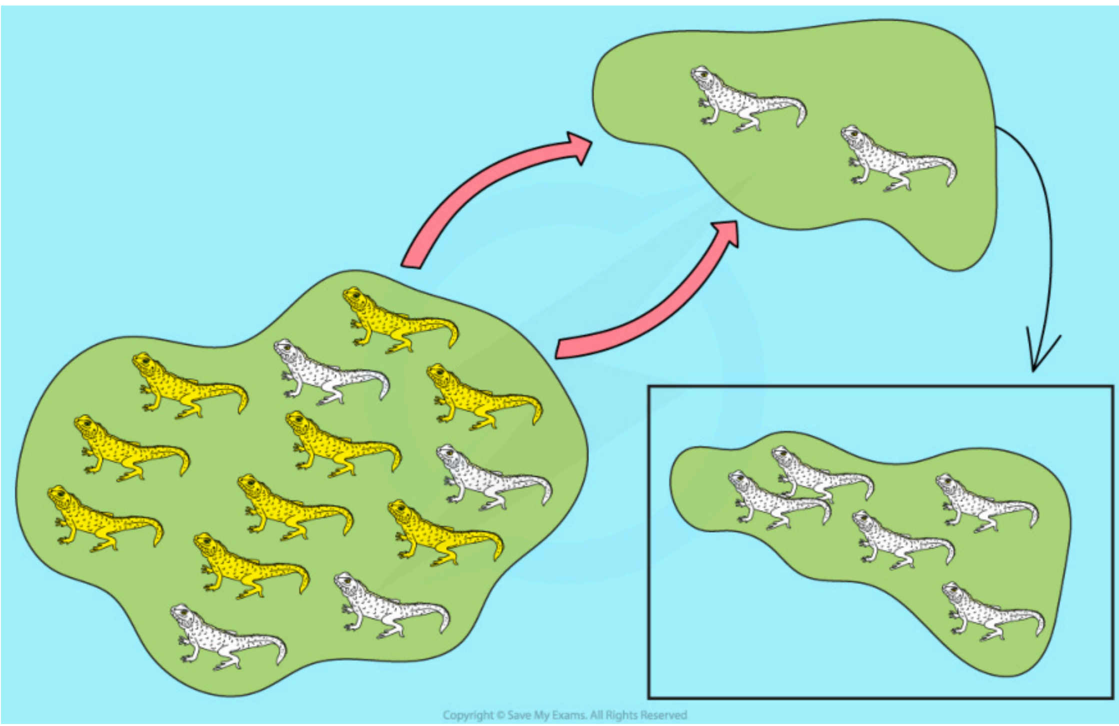
Basis of allele frequency analysis

- For any polymorphism, each individual carries **two alleles per locus**
  - One is inherited from the mother, the other from the father
  - Exception - this does not apply to alleles present on the X or Y chromosome
- Within a population, there are **twice as many total alleles** as there are individuals
- **Homozygous** individuals each contribute **two** of that allele to the total number of that particular allele
- **Heterozygous** individuals each contribute **one** of a particular allele to the total number of that allele
  - For example, if there are eight individuals with the **ZZ** genotype, they contribute 16 **Z** alleles. Thirty-four **Zz** heterozygous individuals contribute a total of 34 **Z** alleles and 34 **z** alleles to the total

Your notes

Worked example

Consider the following isolated island populated by a certain species of lizard. On this island, the ratio of white lizards to yellow lizards is 4:9. The yellow pigmentation is caused by a dominant allele, **Y**. Lizards possessing the homozygous recessive genotype (**yy**) are white in colour.



Distribution of lizards on the island before and after the geological event.

A geological event caused the island to become divided in two; this is shown by the red arrows in the diagram above. One island sank below the ocean, killing all its inhabitants, whilst the other survived. Only two white lizards made it to the surviving half of the island. Fortunately, these were male and female and they were able to begin the recolonisation of the small island.

Calculate the allele frequencies of **Y** and **y** before and after the geological event.

Step 1: Calculate the phenotype frequencies

If 4/13 are the white lizards, the phenotype frequency of **yy** is 4 ÷ 13 = 0.3077

Step 2: Calculate the allele frequency of y (from the phenotype frequency in Step 1)

Your notes

The probability of two y alleles coming together through fertilisation is given by (**q** x **q**) or $\mathbf{q}^2$, where **q** is the allele frequency of the recessive allele, **y**

Therefore, **q** is the square root of the white phenotype frequency

$$q = \sqrt{\frac{4}{13}} = 0.5547$$

Step 3: Subtract this from 1 to find the allele frequency of the dominant allele, Y

1 - 0.5547 = 0.4453

Frequency of **Y** allele before = 0.4453 or 44.5%
Frequency of the **y** allele before = 0.5547 or 55.5%
Frequency of **Y** allele after = 0 or 0%
Frequency of the **y** allele after = 1.0 or 100%

Examiner Tip

Mathematical derivations of allele frequencies are not required for your exams, although it helps to appreciate that the sum total of all the allele frequencies must add up to 1, in order to appreciate the variation within a species.