DP IB Biology: HL

Your notes

10.2 Inheritance

Contents

10.2.1 Unlinked Genes

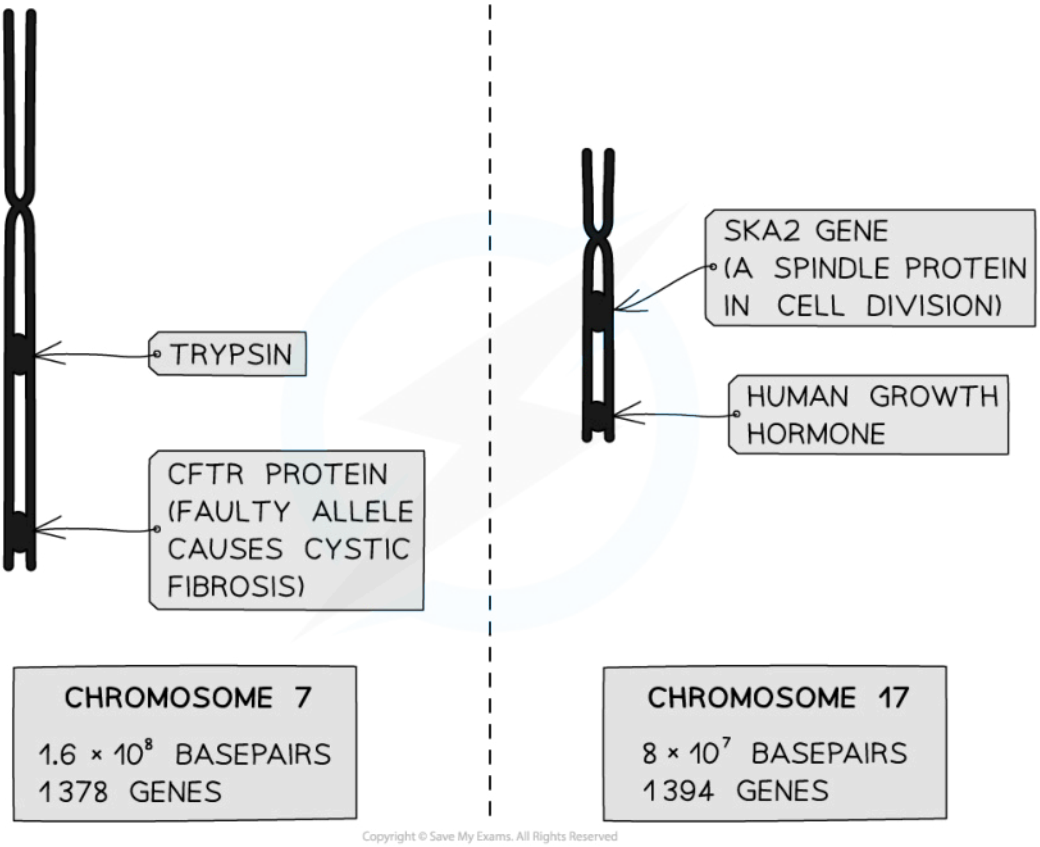

Your notes

Independent Assortment & Segregation

Unlinked genes segregate independently as a result of meiosis

- **Unlinked genes** are genes that an organism carries on **separate chromosomes**
  - Not on homologous copies of the same chromosome
- An example of a pair of unlinked genes in fruit flies (*Drosophila melanogaster)* is
  - The gene for curly wings on **chromosome 2**, and
  - The gene for mahogany eyes on **chromosome 3**
- An example of a pair of unlinked genes in humans is
  - The gene for trypsin (a stomach enzyme) on chromosome **7**, **and**
  - The gene for human growth hormone on chromosome **17**
- **Assortment** of chromosomes refers to their alignment in metaphase I of meiosis
  - Each bivalent assorts (aligns) itself independently of all the others
- **Segregation** of chromosomes (ie. how they get separated) is governed by their pattern of assortment
  - Segregation just refers to **which pole of the cell** the whole chromosomes are pulled to in anaphase I
  - Segregation determines **which combinations of alleles** end up in which gamete cells by the end of meiosis II
- By contrast, linked genes (on the same chromosome) tend to be **inherited together**

Your notes



The loci of selected genes in the human genome

Trypsin and CFTR are linked genes (both on the same chromosome);

Human Growth Hormone and trypsin are unlinked genes (both on different chromosomes)

Punnett Squares for Dihybrid Traits


Your notes

- **Mono**hybrid crosses look at how the alleles of **one** gene transfer across generations
- **Di**hybrid crosses look at how the alleles of **two** genes transfer across generations
  - ie. dihybrid crosses can be used to show the **inheritance** of **two completely different characteristics** in an individual
- The genetic diagrams for both types of cross are very similar
- For dihybrid crosses, there are several more genotypes and phenotypes involved
- When writing out the different genotypes, write the **two alleles for one gene**, followed immediately by the **two alleles for the other gene**.
- Do not mix up the alleles from the different genes
  - For example, if there was a gene with alleles ***Y*** and ***y*** and another gene with alleles ***G*** and ***g*** an example genotype for an individual would be ***YyGg***
- Alleles are usually shown side by side in dihybrid crosses e.g. ***TtBb***

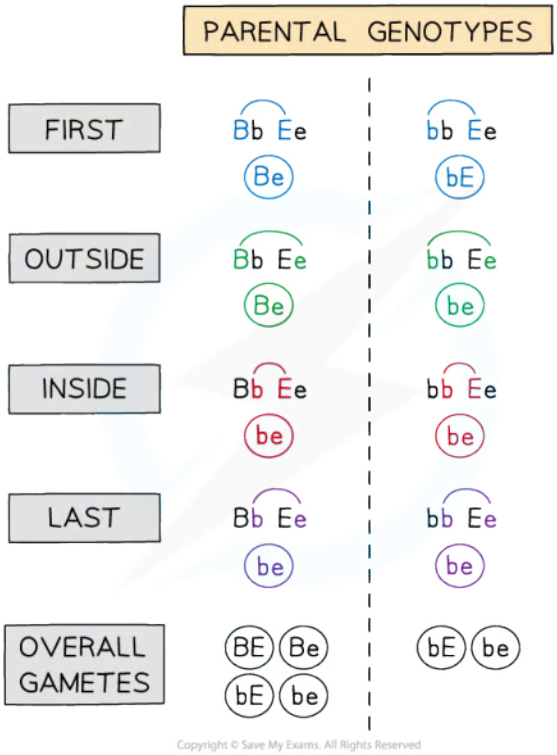
Your notes

Worked example

Worked example 1: Dihybrid genetic diagram

- Horses have a single gene for coat colour that has two alleles:
  - ***B***, a dominant allele produces a black coat
  - ***b***, a recessive allele produces a chestnut coat
- Horses also have a single gene for eye colour
  - ***E***, a dominant allele produces brown eyes
  - ***e***, a recessive allele produces blue eyes
- Each of these genes (consisting of a pair of alleles) are **inherited independently** of one another because the two genes are located on different non-homologous chromosomes
  - Such characteristics are said to be unlinked
- In this example, a horse that is heterozygous for both genes has been crossed with a horse that is homozygous for one gene and heterozygous for the other

Parental phenotypes:	black coat, brown eyes	x	chestnut coat, brown eyes
Parental genotypes:	***BbEe***	x	***bbEe***
Parental gametes:	***BE*** or ***Be*** or ***bE*** or ***be***	x	***bE*** or ***be***



Determining the Alleles Carried by Gametes Based on the Parental Genotypes Using the FOIL (First, Outside, Inside, Last) Method

Your notes

Dihybrid Cross Punnett Square Table

		Gametes from Parent Two	
		bE	be
Gametes from Parent One	BE	BbEE / black coat, brown eyes	BbEe / black coat, brown eyes
	Be	BbEe / black coat, brown eyes	Bbee / black coat, blue eyes
	bE	bbEE / chestnut coat, brown eyes	bbEe / chestnut coat, brown eyes
	be	bbEe / chestnut coat, brown eyes	bbee / chestnut coat, blue eyes

- Predicted ratio of phenotypes in offspring =
  - **3** black coat, brown eyes :
  - **3** chestnut coat, brown eyes :
  - **1** black coat, blue eyes :
  - **1** chestnut coat, blue eyes
- Predicted ratio of genotypes in offspring = **3** ***BbEE*** **:** **3** ***bbEE*** **:** **1** ***Bbee*** **:** **1** ***bbee***

Your notes

Worked example

Worked example 2: Dihybrid genetic diagram

In a separate cross to that shown in Worked Example 1, a horse that is heterozygous for both genes has been crossed with another horse that is heterozygous for both genes.

Parental phenotypes:	black coat, brown eyes	x	black coat, brown eyes
Parental genotypes:	***BbEe***	x	***BbEe***
Parental gametes:	***BE*** or ***Be*** or ***bE*** or ***be***	x	***BE*** or ***Be*** or ***bE*** or ***be***

Dihybrid Cross Punnett Square Table 2

		Gametes from Parent Two			
		BE	Be	bE	be
Gametes from Parent One	BE	BBEE / black coat, brown eyes	BBEe / black coat, brown eyes	BbEE / black coat, brown eyes	BbEe / black coat, brown eyes
	Be	BBEe / black coat, brown eyes	BBee / black coat, blue eyes	BbEe / black coat, brown eyes	Bbee / black coat, blue eyes
	bE	BbEE / black coat, brown eyes	BbEe / black coat, brown eyes	bbEE / chestnut coat, brown eyes	bbEe / chestnut coat, brown eyes
	be	BbEe / black coat, brown eyes	Bbee / black coat, blue eyes	bbEe / chestnut coat, brown eyes	bbee / chestnut coat, blue eyes

- Predicted ratio of phenotypes in offspring =
  - **9** black coat, brown eyes :
  - **3** chestnut coat, brown eyes :
  - **3** black coat, blue eyes :
  - **1** chestnut coat, blue eyes

Examiner Tip

For the double-heterozygous cross for unlinked genes above, you're expected to remember the phenotypic ratio 9:3:3:1. You won't need to remember the ratio of the genotypes but this can be worked out from a Punnett square like the one above.

10.2.2 Skills: Analysing Dihybrid Crosses

Your notes

Predicting Phenotypic and Genotypic Ratios

Worked example

Fruit flies (*Drosophila melanogaster*) were crossed in a laboratory study looking for inheritance patterns for two characteristics, wing length and body colour. We can assume that these characteristics are unlinked.

The alleles for these characteristics are as follows:

V = long wings

v = short (vestigial) wings

B = brown body colour

b = black body colour

A black-bodied, heterozygous long-winged fly was crossed with short-winged, homozygous brown-bodied flies. Predict the phenotype ratio of their offspring.

Step 1: Write out the parental genotypes

The first parent is black-bodied and heterozygous long-winged. To be black-bodied it must have the genotype ***bb***. Heterozygous long-winged has the genotype ***Vv***
The second parent is short-winged and homozygous brown-bodied. To be short-winged it must have the genotype ***vv***. Homozygous brown-bodied has the genotype ***BB***

Vvbb × ***vvBB***

Step 2: Identify the gamete genotypes that each parent could produce

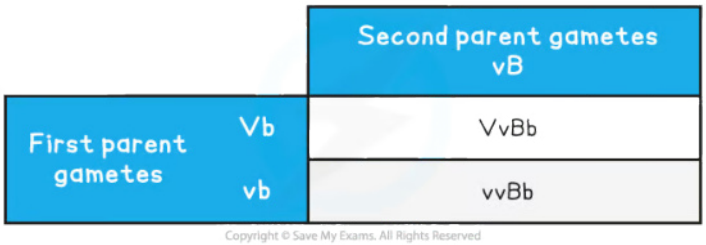
These are the allele combinations that each parent can produce in meiosis

Vb ***vb*** × ***vB***

Step 3: Complete a Punnett square to show the genotypes of the offspring

Punnett Square showing Genotypes of the Offspring

Your notes



Step 4: Identify the phenotypes of the offspring

VvBb = long-winged, brown-bodied
vvBb = short-winged, brown bodied

Conclusion: The offspring would be 100% brown-bodied and 1:1 long to short-winged

Test crosses

- A test cross can be used to **deduce the genotype**
- The individual in question is crossed with an individual that is expressing the **recessive phenotype**
- This is because an individual with a recessive phenotype has a known genotype
- The resulting phenotypes of the offspring provide sufficient information to suggest the genotype of the unknown individual
- For a monohybrid test cross:
  - If **no** offspring exhibit the recessive phenotype then the unknown genotype is **homozygous dominant**
  - If **at least one** of the offspring exhibit the recessive phenotype then the unknown genotype is **heterozygous**
- For a dihybrid test cross:
  - If no offspring exhibit the recessive phenotype for either gene then the unknown genotype is **homozygous dominant** for both genes
  - If at least one of the offspring exhibit the recessive phenotype for one gene but not the other, then the unknown genotype is **heterozygous for one gene and homozygous dominant for the other**
  - If at least one of the offspring exhibit the recessive phenotype for both genes then the unknown genotype is **heterozygous for both** genes

Your notes

Worked example

Worked example: Test crosses

- Rabbits have a single gene for ear length that has two alleles:
  - ***D***, a dominant allele that produces long ears
  - ***d***, a recessive allele that produces shorter ears
- A breeder has a rabbit called Floppy that has long ears and they want to know the genotype of the rabbit
  - There are two possibilities: ***DD*** or ***Dd***
- The breeder crosses the long-eared rabbit with a short-eared rabbit
  - A rabbit displaying the recessive short ear phenotype has to have the genotype ***dd***

Test Cross Possibility Table

		Known gametes	
		d	d
Possible gametes Option 1	D	Dd / long ears	Dd / long ears
	D	Dd / long ears	Dd / long ears

- The predicted ratio of phenotypes of offspring – 1 long ears
- The predicted ratio of genotypes of offspring – 1 Dd

Your notes

Test Cross Possibility Two Table

		Known gametes	
		d	d
Possible gametes Option 2	D	Dd / long ears	Dd / long ears
	d	dd / short ears	dd / short ears

- Predicted ratio of phenotypes of offspring – 1 long ears : 1 short ears
- Predicted ratio of genotypes of offspring – 1 *Dd* : 1 *dd*
- The breeder identifies the different phenotypes present in the offspring
- There is at least one offspring with the short ear phenotype
- This tells the breeder that their rabbit Floppy has the genotype ***Dd***
- If Floppy was genotype ***DD*** none of the offspring would have short ears

Your notes

Worked example

Worked example: Hard Question

A farmer wishes to maximise his yield of soybean oil from his crop. Oil is extracted from the seeds which typically measure 6–12 mm in diameter. In one species of soybean, Glycine max, two characteristics are governed by the following pairs of **unlinked** alleles.
H = high oil content in the seeds; ***h*** = low oil content in the seeds
E = four seeds in a pod; ***e*** = two seeds in a pod
The farmer crossed two soybean plants, both with high oil content and four seeds per pod. This cross resulted in 381 offspring plants being produced. The 381 F_1 offspring had a phenotypic spread as shown in the table below.

Phenotype of F_1 generation		Numbers of plants in F_1 generation
Oil Content	Number of Seeds per Pod	
High	4	215
High	2	73
Low	4	70
Low	2	23
		381

Use the data in the table to deduce the genotypes of the parent plants in this cross.

Step 1: Write out the possible parental genotypes

Both parents were high-oil content with 4 seeds per pod, which are the dominant traits for each pair of alleles. Neither parent can be homozygous recessive for either oil content or the number of seeds.

Possible allele combinations for oil content: ***HH***, ***Hh***
Possible allele combinations for 4 seeds per pod: ***EE***, ***Ee***
Therefore, the possible parental genotypes were: ***HHEE***, ***HHEe***, ***HhEE***, ***HhEe***

Step 2: Examine phenotype ratios

The smallest number of F_1 offspring was 23 for low oil content, 2 seeds. These must have the homozygous recessive genotype, ***hhee***.

The ratios of other phenotypes, relative to the homozygous recessive phenotype, were:
low oil, 4 seeds : ***hhee*** = 70:23 = 3.04 : 1 ≅ 3:1
high oil, 2 seeds : ***hhee*** = 73:23 = 3.17 : 1 ≅ 3:1
high oil, 4 seeds : ***hhee*** = 215:23 = 9.3 : 1 ≅ 9:1

Step 3: Apply Mendel's Law of independent assortment for linked genes

The phenotypes of the offspring display an approximate 9:3:3:1 ratio. This characterises a cross between two double-heterozygous parents where the genes are unlinked, as in this case

Conclusion: Both parents' genotype was ***HhEe*** / double heterozygous

Examiner Tip

Make sure before you start a test cross you think about the following: how many genes are there, how many alleles of each gene are there, which is the dominant allele, what type of dominance is it and is there linkage or codominance between genes?

Even though we learn about fruit flies (*Drosophila melanogaster*) being studied in detail which led to the discovery of gene linkage, many of their characteristics are unlinked and can be studied on a large scale in this manner.

Your notes

10.2.3 Gene Linkage


Your notes

Exceptions to Mendel's Rules

NOS: Mendel used observations of the natural world to find and explain patterns and trends

- Since Mendel, scientists have looked for discrepancies and asked questions based on further observations to show exceptions to the rules. For example, Morgan discovered non-Mendelian ratios in his experiments with *Drosophila*
- When looking at dihybrid crosses (crosses with two pairs of observable characteristics), Mendel explained his experimental data with his **law of independent assortment**
- That individual characteristics are **inherited completely independently** of each other
- In many cases, **this is correct**
- The significance of Mendel's work went **largely unnoticed for decades**, until after his death in 1884
- Scientists in the 1890s and early 1900s picked up his experimental findings and **replicated them**
- The **large number of trials** that Mendel undertook highlighted patterns/trends in the inheritance of certain factors (what are now known as genes)
- However, **discrepancies were noticed** when scientists replicated Mendel's experiments
  - And also when experiments were undertaken on the inheritance of certain genes in **other organisms**
- Many of their dihybrid crosses replicated the **9:3:3:1 pattern of phenotypes** that Mendel first observed in his work
- **William Bateson** and **Reginald Punnett**, two Cambridge University geneticists (in collaboration with a third Cambridge biologist, **Edith Saunders**), replicated Mendel's findings, again in experiments with sweet peas, however
- They discovered some **apparently anomalous results** in certain cases, in which phenotype ratios **did not follow** the classical 9:3:3:1 pattern
- Many scientists **would have dismissed non-conforming** results as mere anomalies, however Bateson, Punnett and Saunders chose to search for an explanation
- Punnett's quote from one his **laboratory notebooks** sums up their approach:
  - "*Treasure your exceptions! When there are none, the work gets so dull that no one cares to carry it further.*"
- Bateson and Punnett performed further work, mainly on crossings of sweet peas and crossings of chickens, but were **unable to offer a robust explanation** for certain unpredictable phenotype ratios in their crosses

Gene Linkage

Your notes

Gene loci are said to be linked if they are on the same chromosome

- **Loci** (singular: locus) refers to the **specific linear positions** on the chromosome that genes occupy
- If genes are on the sex chromosome, they are said to be **sex-linked**
  - Sex-linked genes have characteristics that generally **only affect one gender** of a species
  - These genes are usually **on the X chromosome** because the Y chromosome contains very few genes
  - In humans, **colour-blindness** and **haemophilia** are notable examples of genetic conditions that only affect males
- Linked genes located on the chromosomes 1–22, or any chromosome that is not a sex chromosome (called autosomes) are said to be examples of **autosomal linkage**
- The likelihood of genes being inherited together, or the extent to which they are linked, is measured in units called **centimorgans**, in honour of Thomas Hunt Morgan's work

Notation for link genes

- When writing linked genotypes it can be easier to keep the linked alleles within a bracket
  - For example, an individual has the genotype ***FFGG***. However, if there is linkage between the two genes, it would be written as (***FG***)(***FG***)
- Another commonly-used way of denoting linked alleles is to link them with a line. So, for example, linkage between genes ***F*** and ***G*** might be shown as

F ***G***

_____

_____

f ***g***

- Remember to distinguish between sex linkage and autosomal linkage. The explanation of non-Mendelian ratios falls into the domain of autosomal linkage for IB

Autosomal linkage

- Dihybrid crosses and their predictions rely on the assumption that the genes being investigated behave **independently of one another** during meiosis
- However, **not all genes assort independently** during meiosis
- Some genes which are located on the **same chromosome** display autosomal **linkage** and **stay together in the original parental combination**
- Linkage between genes affects how parental alleles are passed onto offspring through the gametes

Identifying autosomal linkage from phenotypic ratios

- In the following **theoretical example**, a dihybrid cross is used to predict the inheritance of **two different characteristics** in a species of newt
  - The genes are for **tail length** and **scale colour**
- The gene for tail length has two alleles:
  - Dominant allele **T** produces a normal length tail

- Recessive allele **t** produces a shorter length tail
- The gene for scale colour has two alleles:
  - Dominant allele **G** produces green scales
  - Recessive allele **g** produces white scales

Your notes

Without linkage

- The outcomes for this dihybrid cross if the genes are **unlinked** are as follows

Dihybrid Cross without Linkage Punnett Square Table

		Gametes from Parent Two: tg
Gametes from Parent One	TG	TtGg / normal tail, green scales
	Tg	Ttgg / normal tail, white scales
	tG	ttGg / short tail, green scales
	tg	ttgg / short tail, white scales

- Predicted ratio of phenotypes in offspring =
  - 1 normal tail, green scales : 1 normal tail, white scales : 1 short tail, green scales : 1 short tail, white scales
- Predicted ratio of genotypes in offspring =
  - 1 TtGg : 1 Ttgg : 1 ttGg : 1 ttgg

With linkage

- However, if the **same dihybrid cross** is carried out but this time the genes are **linked**, we get a **different phenotypic ratio**
  - There would be a **1** : **1** phenotypic ratio (**1** normal tail, green scales : **1** short tail, white scales)

- This change in the phenotypic ratio occurs because the genes are located on the **same chromosome**
- The **unexpected phenotypic ratio**, therefore, shows us that the genes are **linked**

- The explanation for this new phenotypic ratio is given in the worked example below:

Your notes

Worked example

Worked example: Explaining autosomal linkage

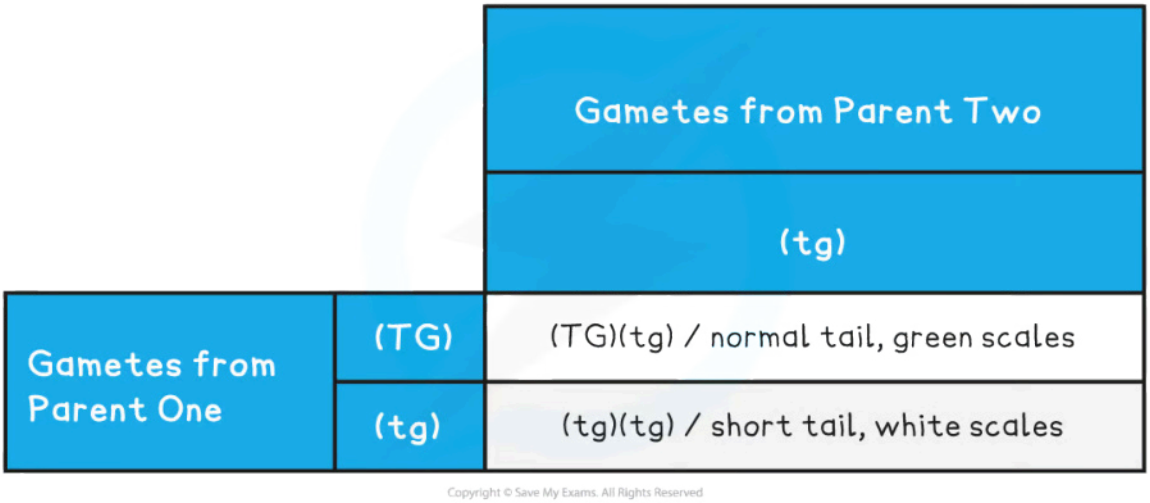
- In reality, the genes for tail length and scale colour in this particular species of newt show autosomal linkage

Parental phenotypes: normal tail, green scales x short tail, white scales

Parental genotypes: **(TG)(tg)** **(tg)(tg)**

Parental gametes: **(TG)** or **(tg)** **(tg)**

Dihybrid Cross with Linkage Punnett Square Table



- Predicted ratio of genotypes in offspring =
  - **1** (TG)(tg) : **1** (tg)(tg)
- Predicted ratio of phenotypes in offspring =
  - **1** normal tail, green scales : **1** short tail, white scales

Examiner Tip

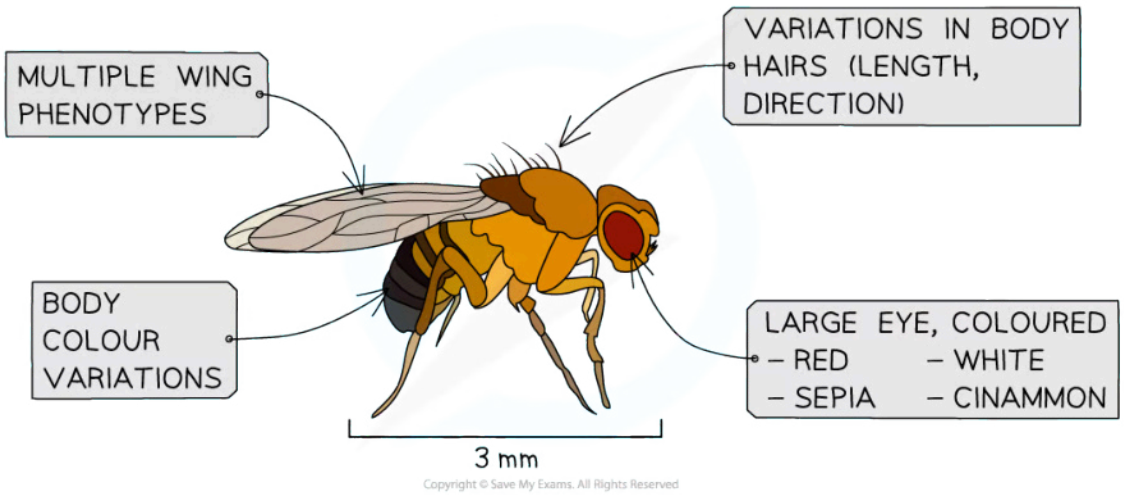
When you are working through different genetics questions you may notice that test crosses involving autosomal linkage predict solely **parental type** offspring (offspring that have the same combination of characteristics as their parents). However in reality **recombinant** offspring (offspring that have a different combination of characteristics to their parents) are often produced. This is due to the **crossing over** that occurs during meiosis. The crossing over and exchanging of genetic material **breaks the linkage** between the genes and recombines the characteristics of the parents. So if a question comes along that asks you why recombinant offspring are present you now know why!

Your notes

Non-Mendelian Ratios in Drosophila

Your notes

- In the way that Bateson, Punnett and Saunders developed and refined Mendel's findings, Thomas Hunt Morgan further refined genetic theory
  - His work was awarded the Nobel Prize in 1933

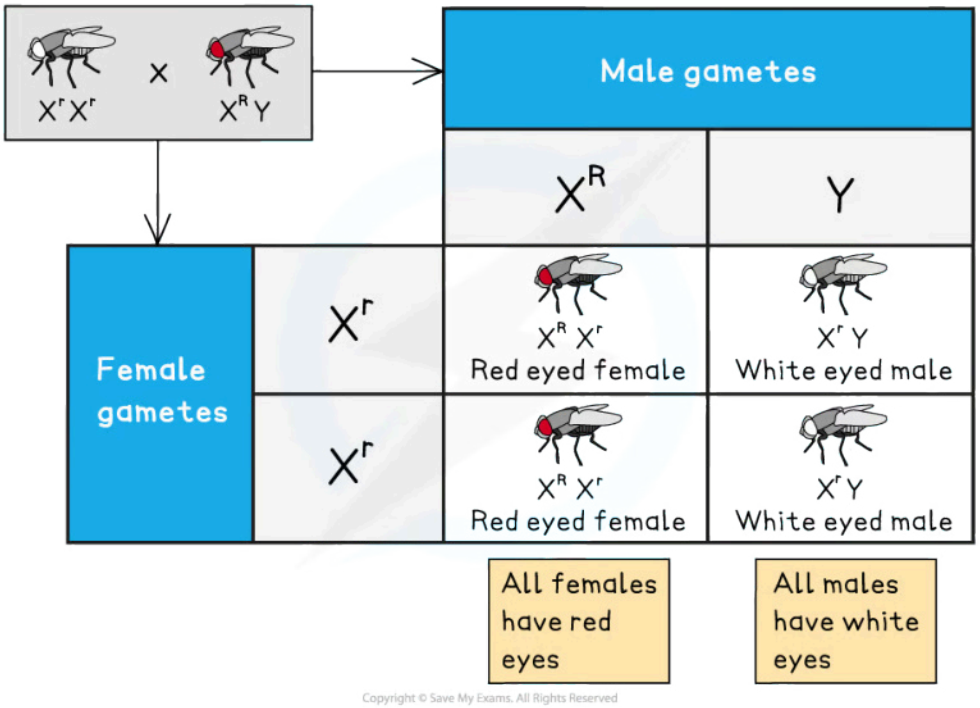


A fruit fly* (Drosophila melanogaster) *with some of the phenotypic variations observed in Thomas Hunt Morgan's work

Sex linkage

- Working in the USA in the early 20th century, he bred fruit flies (Drosophila melanogaster) over successive generations
- In his cross-breeding experiments he came across red-eyed wild types and white-eyed mutants
- He realised there was a distinct **sex bias in phenotypic distribution**
  - All-female offspring of a red-eyed male were red-eyed while all male offspring of a white-eyed female were also white-eyed
- Morgan hypothesised that this occurred because the **gene for eye colour** was located on a **sex chromosome** (i.e. X-linked)

Your notes



Sex linkage in Drosophila. A cross between a homozygous white-eyed female and a male with red eyes gives all white-eyed males and red-eyed female offspring

Autosomal linkage

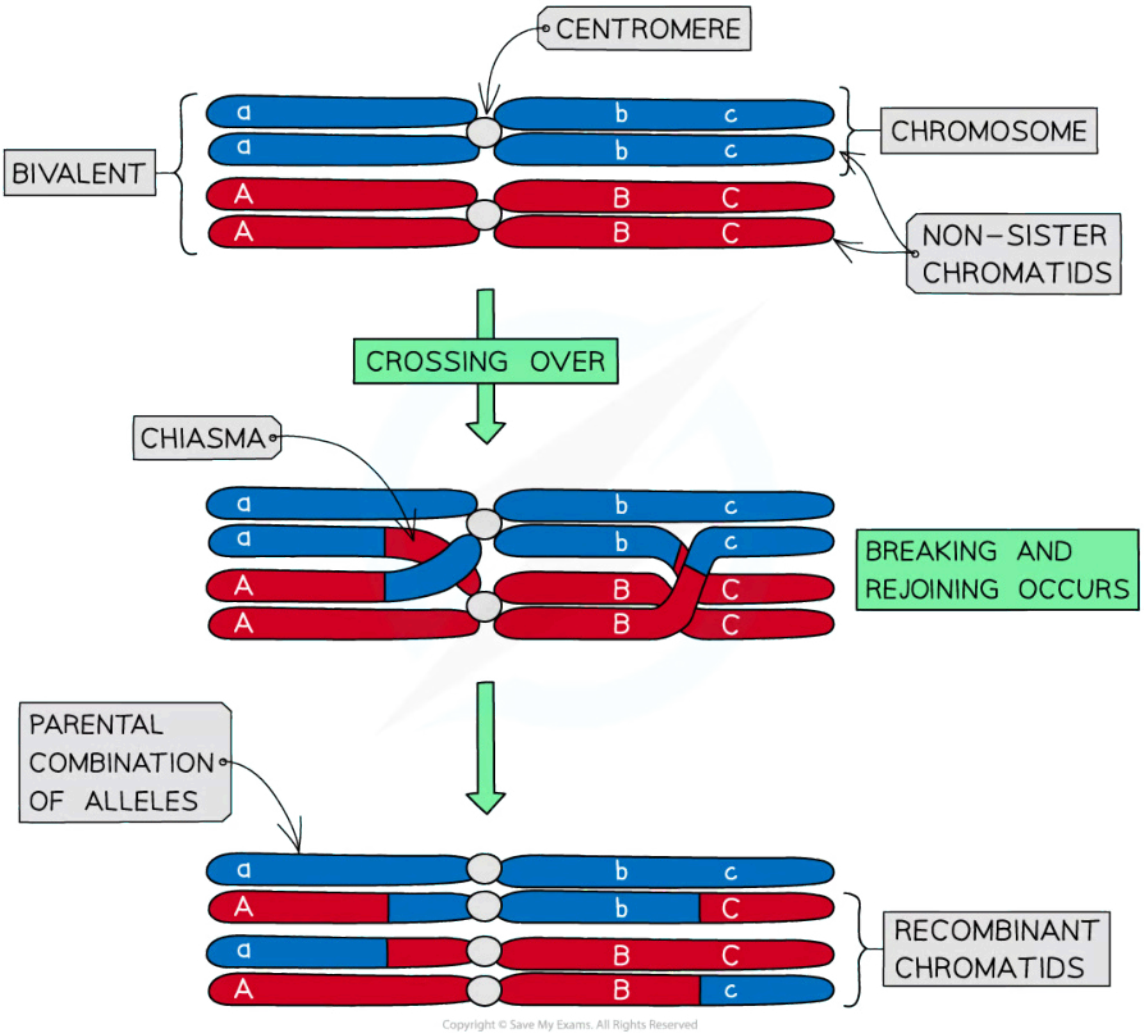
- As Morgan continued his experiments he noticed a number of different traits in fruit flies that did not conform to Mendelian ratios as **several phenotypes occurred in much lower frequencies** than expected
- Based on this data, Morgan made two key proposals:
  - The alleles for these traits were located on **the same chromosom**e (gene linkage) meaning they did **not independently assort**
  - Linked alleles could be **unlinked via recombination** (crossing over) to produce **recombinant** offspring (offspring that have a different combination of characteristics to their parents)
    - This is due to the **crossing over** that occurs during meiosis
    - The crossing over and exchanging of genetic material **breaks the linkage** between the genes and recombines the characteristics of the parents
- Morgan also observed that the number of recombinants that resulted from crossing linked genes **varied** depending on the combination of traits
- He proposed the idea that the number of recombinants (**crossover frequency)** may be related to the **distance between two genes** on the same chromosome
  - Genes that were further apart had a **higher crossover frequency,** whereas genes closer together exhibited a lower crossover frequency
  - Morgan used this concept to create the first **gene linkage maps**
    - These maps displayed the **relative positions of genes** on chromosomes

10.2.4 Skills: Identifying Recombinants

Your notes

Identifying Recombinants in Crosses

- Genetic diagrams involving autosomal linkage often predict solely **parental type** offspring (offspring that have the same combination of characteristics as their parents)
- However in reality **recombinant** offspring (offspring that have a different combination of characteristics to their parents) are often produced
  - This is due to the **crossing over** that occurs during meiosis
  - The crossing over and exchanging of genetic material **breaks the linkage** between the genes and recombines the characteristics of the parents



The process of crossing-over results in recombinant phenotypes that can differ from the parental phenotype.

- The frequency of recombinants within a population will nearly always be less than that of non-recombinants
  - Crossing over is **random** and chiasmata form at different locations with each meiotic division
- **Recombination frequency** between two linked genes is **greater when genes are further apart** on the same chromosome
  - There are more possible locations for a chiasma to form between the genes


Your notes

Identifying recombinants using test crosses

- **Test crosses** are often used to determine unknown genotypes
- Similarly, they can be used to **identify recombinant phenotypes in offspring**
- An individual is crossed with a **homozygous recessive individual (for both traits)**
  - If **any** of the offspring possess a **non-parental phenotype** then they are labelled as **recombinants**
    - These individuals have **new allele combinations** due to the process of crossing over during meiosis leading to the exchange of genetic material between chromosomes

Drawing a Punnett square to show dihybrid inheritance of linked genes

- A number of sweet pea plants were generated by crossing double-homozygous dominant plants (***PL***)(***PL***) with double-homozygous recessive plants (***pl***)(***pl***) to produce a 100% heterozygous F_1 generation (***PL***)(***pl***) as expected
- Members of this generation were then interbred to produce the F_2 generation
- Alleles:
  - ***P*** = purple flowers, dominant to ***p*** = red flowers
  - ***L*** = long seeds, dominant to ***l*** = round seeds

Possible Gametes Table

Your notes

Grandparent of F2 generation – genotypes	$\frac{PL}{PL}$ x $\frac{pl}{pl}$
Gametes	PL pl
F1 generation genotype	$\frac{PL}{pl}$
Due to crossing over, the F1 generation will produce some recombinant gametes as well as a majority of parental ones	
F1 x F2 cross – genotypes	$\frac{PL}{pl}$ x $\frac{PL}{pl}$
Gametes – parental These will be abundant	PL pl PL pl
Gametes – recombinant These will be scarce	pL Pl pL Pl

F_2 Punnet Square Showing Possible Genotypes

		Gametes from F_1 generation, parent 1			
		P L	p l	p L	P l
Gametes from F_1 generation, parent 2	P L	$\frac{PL}{PL}$	$\frac{PL}{pl}$	$\frac{PL}{pL}$	$\frac{PL}{Pl}$
	p l	$\frac{pl}{PL}$	$\frac{pl}{pl}$	$\frac{pl}{pL}$	$\frac{pl}{Pl}$
	p L	$\frac{pL}{PL}$	$\frac{pL}{pl}$	$\frac{pL}{pL}$	$\frac{pL}{Pl}$
	P l	$\frac{Pl}{PL}$	$\frac{Pl}{pl}$	$\frac{Pl}{pL}$	$\frac{Pl}{Pl}$

- According to Mendelian ratios and the Punnett square, the F_2 generation should follow the typical 9:3:3:1 ratio

Your notes

- However, in reality, the **frequency of recombinant gametes will be much lower** than that of parental gametes
  - This affects the resulting offspring phenotypes, with **fewer recombinant phenotypes occurring** than expected

Expected vs Predicted Phenotypes Table

EXPECTED phenotype % in F_2 offspring (9 : 3 : 3 : 1 ratio)	ACTUAL phenotype % in F_2 offspring	Phenotype	Observations
56%	70%	Purple flower, long pollen grains	More than expected
19%	5%	Purple flower, round pollen grains	Less than expected
19%	6%	Red flower, long pollen grains	Less than expected
6%	19%	Red flower, round pollen grains	More than expected

Observations

- More of the F_2 offspring than expected showed the **parental phenotypes**
- **Fewer** plants with **recombinant phenotypes** were produced than the 9:3:3:1 ratio would suggest
- The actual ratios found were referred to as '**non-Mendelian**' as they didn't follow Mendel's pattern
- However, this was not zero; **some recombinants** were still being produced

Possible Theories to Explain These Findings

- At the time, it was known that **many genes were carried on a few chromosomes**
- The idea that certain genes **share the same chromosome** was being developed by many scientists
- This suggested that genes **could be inherited together**, not by the law of independent assortment as put forward by Mendel
- The idea of linkage of genes was developed to explain the non-Mendelian ratios
  - The frequency of recombinant phenotypes is **lower** because **crossing over is a random process** and the chiasmata do not always form in the same place for each meiotic division
  - The frequency of recombinant gametes also depends on the **closeness of linkage** between the two genes
    - Genes located **close together** on a chromosome are **less likely** to be separated by crossing over
    - So recombinants of those two genes will be less frequent
- Thomas Hunt Morgan later provided proof of linkage to explain non-Mendelian ratios in his experimentation with fruit flies (Drosophila melanogaster)

Examiner Tip

Remember to distinguish between sex linkage and autosomal linkage. The explanation of non-Mendelian ratios falls into the domain of autosomal linkage for IB.

Your notes

10.2.5 Skills: Chi-squared Test

Your notes

Chi-squared Test and Dihybrid Crosses

Use of a chi-squared test on data from dihybrid crosses

- The difference between **expected** and **observed** results in experiments can be statistically significant or insignificant (happened by chance)
- If the difference between results is statistically significant it can suggest that something else is happening in the experiment that isn't being accounted for
  - For example, linkage between genes
- A statistical test called the chi-squared test determines whether there is a **significant difference** between the observed and expected results in an experiment
- The chi-squared test is completed when the data is **categorical** (data that can be grouped)

Calculating chi-squared values

- Obtain the expected and observed results for the experiment
- Calculate the difference between each set of results
- Square each difference (as it is irrelevant whether the difference is positive or negative)
- Divide each squared difference by the expected value and get a sum of these answers to obtain the chi-squared value

THE CHI-SQUARED VALUE, χ^2 IS GIVEN BY THE FORMULA

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

Σ = SUM OF

O = OBSERVED VALUE

E = EXPECTED VALUE

Analysing chi-squared values

- To work out what the chi-squared value means, **a table that relates chi-squared values to probabilities** is used
- If the chi-squared value represents a **larger probability than the critical probability** then it can be stated that the differences between the expected and observed results are **due to chance**
- If it represents a **smaller probability than the critical probability** then the differences in results are **significant** and something else may be causing the differences
- To determine the critical probability biologists generally use a probability of **0.05** (they allow that chance will cause five out of every 100 experiments to be different)

- The number of comparisons made must also be taken into account when determining the critical probability. This is known as the **degrees of freedom**

Your notes

Worked example

An experiment was carried out investigating the inheritance of two genes in rabbits

- One for coat colour and one for ear length
- A dihybrid cross revealed the expected ratio of phenotypes to be 9 : 3 : 3 : 1
- Several rabbits with the double-heterozygous genotype were bred together and the phenotypes of all the offspring were recorded
- The ratio of the offspring was **not exactly what was predicted**, but was reasonably close
- In order to determine whether this was due to chance or for some other reason, the chi-squared test was used

Chi-squared Worked Example Table 1

Phenotypes & (genotypes) of F_2 offspring		Observed Number (O)	Expected Ratio	Expected Number (E)	O – E	$(O - E)^2$	$(O - E)^2 / E$
Brown coat (BB / Bb)	Long ears (EE / Ee)	87	9 : 3 : 3 : 1	90	−3	9	0.1000
Brown coat (BB / Bb)	Short ears (ee)	31		30	1	1	0.0333
Black coat (bb)	Long ears (EE / Ee)	27		30	−3	9	0.3000
Black coat (bb)	Short ears (ee)	15		10	5	25	2.5000
		160		160		$\Sigma =$	2.9333
						$\chi^2 =$	2.93

- The expected number of each phenotype is the fraction of the total number of rabbits governed by the 9:3:3:1 ratio
- These are $^9/_{16}$, $^3/_{16}$, $^3/_{16}$ and $^1/_{16}$ of 160, respectively
- In order to understand what this chi-squared value of 2.93 says about the data, a table relating chi-squared values to probability is needed

Relating Chi-Squared Values to Probability Table

Your notes

Degrees of freedom	Probability that the difference between observed and expected results is due to chance			
	0.1	0.05	0.01	0.001
1	2.71	3.84	6.64	10.83
2	4.60	5.99	9.21	13.82
3	6.25	7.82	11.34	16.27
4	7.78	9.49	13.28	18.46

- The chi-squared table displays the probabilities that the differences between expected and observed are **due to chance**
- The **degrees of freedom** can be worked out from the results. It is calculated by **subtracting one from the number of classes**
- In this example, there are four phenotypes which means four classes, 4 – 1 = 3
- This means that the values in the **third row** are important for comparison
- For this experiment, there is a **critical probability of 0.05**
- This means that **7.82** is the value used for comparison
- The chi-squared value from the results (2.93) is **much smaller than 7.82**
- 2.93 would be located somewhere to the left-hand side of the table, representing a probability much greater than 0.1
- This means that there is **no significant difference** between the expected and observed results, any differences that do occur are **due to chance**

Your notes

Worked example

An experiment was carried out on *Drosophila* genes

- One gene for body colour ***B*** = brown, ***b*** = black
- One gene for wing shape ***W*** = straight, ***w*** = curved
- A dihybrid cross revealed the expected ratio of phenotypes to be 9 : 3 : 3 : 1
- Several *Drosophila* with the double-heterozygous genotype were bred together and the phenotypes of all the F_2 offspring were recorded
- The ratio of the offspring was **far from what was predicted**
- In order to determine whether this was due to chance or for some other reason, the **chi-squared test** was used

Chi-squared Worked Example 2 Table

Phenotypes & (genotypes) of F_2 offspring		Observed Number (O)	Expected Ratio	Expected Number (E)	O – E	$(O - E)^2$	$(O - E)^2 / E$
Brown body (BB / Bb)	Straight wing (WW / Ww)	450	9 : 3 : 3 : 1	336	114	12,996	39
Brown body (BB / Bb)	Curved wing (ww)	31		112	−81	6,561	59
Black body (bb)	Straight wing (WW / Ww)	27		112	−85	7,225	65
Black body (bb)	Curved wing (ww)	89		37	52	2,704	73
		597		597		$\Sigma =$	235
						$\chi^2 =$	235

- The expected number of each phenotype is the fraction of the total number of flies governed by the 9:3:3:1 ratio
- These are $^9/_{16}$, $^3/_{16}$, $^3/_{16}$ and $^1/_{16}$ of 597, respectively
- In order to understand what this chi-squared value of 235 says about the data, a table relating chi-squared values to probability is needed

Relating Chi-Squared Values to Probability Table

Your notes

Degrees of freedom	Probability that the difference between observed and expected results is due to chance			
	0.1	0.05	0.01	0.001
1	2.71	3.84	6.64	10.83
2	4.60	5.99	9.21	13.82
3	6.25	7.82	11.34	16.27
4	7.78	9.49	13.28	18.46

- The chi-squared table displays the probabilities that the differences between expected and observed are **due to chance**
- The **degrees of freedom** can be worked out from the results. It is calculated by **subtracting one from the number of classes**
- In this example, there are also four phenotypes which means four classes, 4 – 1 = 3
- This means that the values in the **third row** are important for comparison
- For this experiment, there is a **critical probability of 0.05**
- This means that **7.82** is the value used for comparison
- The chi-squared value from the results (235) is **much greater than** 7.82
- 235 would be located somewhere off the far right of the table, representing a very **small probability**, much less than 0.001
- This means that there is a **significant difference** between the expected and observed results, any differences that do occur are **due to a factor other than chance**
- In this case, that factor is (autosomal) **gene linkage**

Conclusion

- The alleles for black/brown body colour and straight/curved wings are linked, ie. carried on the same chromosome (autosome)

Examiner Tip

When calculating a chi-squared value it is very helpful to create a table like the ones seen in the worked examples. This will help you with your calculations and make sure you don't get muddled up! You should also be prepared to suggest reasons why results might be significantly different. For example, there could be linkage between the genes being analysed.

Your notes

10.2.6 Variation

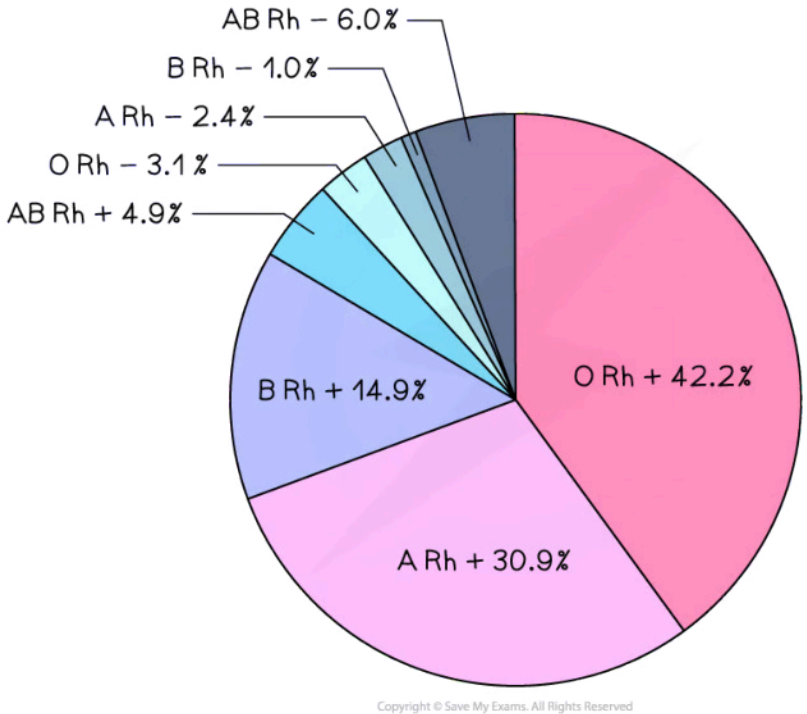
Your notes

Types of Variation

- The ways in which organisms differ from one another is called **variation**
- Variation occurs **between species**
  - In fact, species are classified based on differences between their respective members
  - This is called **interspecific variation**
- Variation can occur **within the same species**
  - Between different individuals or groups of individuals
  - This is called **intraspecific variation**
  - This suggests that only one gene is involved in governing discrete variation
  - This is called monogenic inheritance

Variation can be discrete or continuous

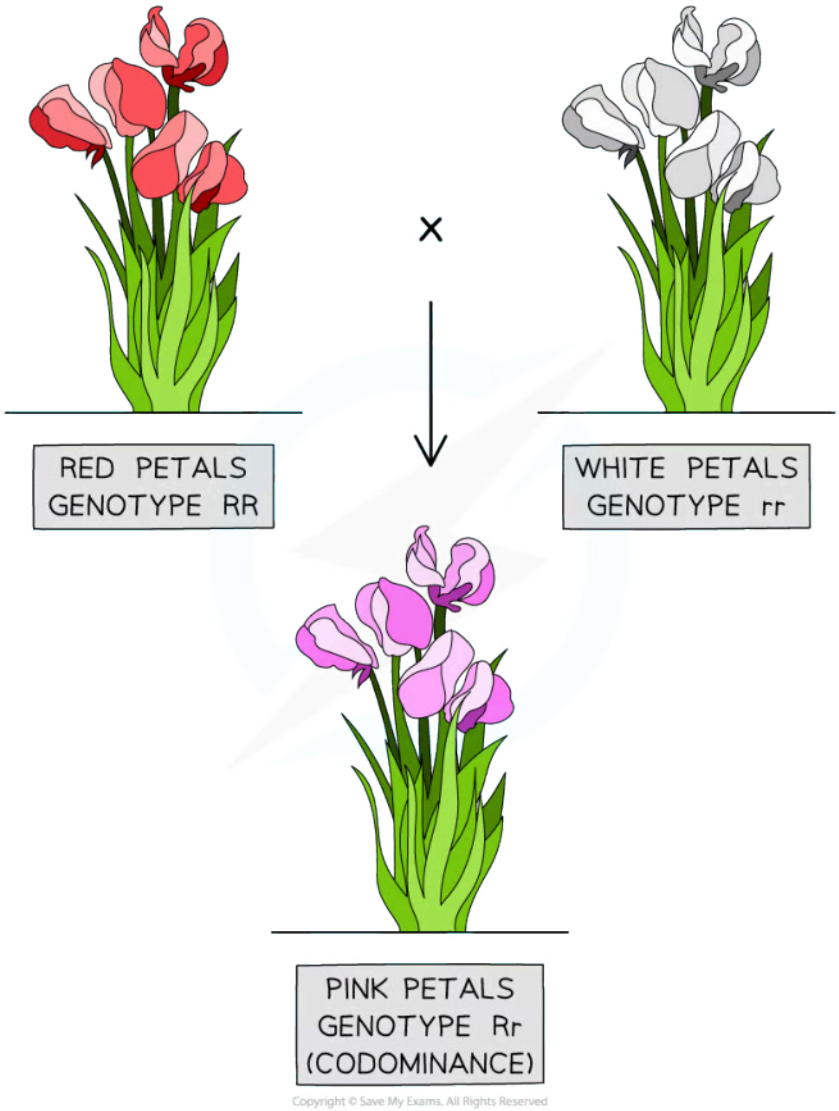
- Discrete variation is an example of intraspecific variation
  - Individuals fall into two or more clear-cut categories with no overlap or in-between categories
  - Blood group is an example of discrete variation
    - All human blood is either group O, A, B or AB, each with a Rhesus factor (+ or -)
- This gives just 8 distinct blood groups



Worldwide A, B, O blood group distribution by percentage, 2019

(data varies regionally with ethnicity)

- The petal colour of snapdragons is a discrete variable; either red, white or pink with no in-between colours
- Discrete variation is sometimes referred to as **discontinuous** variation, in contrast to continuous variation



Snapdragons display 3 main petal colours: red, white and pink, determined by a single pair of codominant alleles

This is an example of discrete variation that is solely due to genetic factors

Causes of discrete variation

Your notes

- This type of variation occurs solely due to **genetic factors**
- The environment has no direct effect
  - Phenotype = **genotype**
- At the genetic level:
  - Different **genes** have **different effects** on the phenotype
  - Different **alleles** at a single gene locus have a **large effect** on the phenotype
  - Remember diploid organisms will inherit two alleles of each gene, these alleles can be the same or different
- A good example of this is the *F8* gene that codes for the blood-clotting protein Factor VIII
  - The different alleles at the *F8* gene locus dictate whether or not normal Factor VIII is produced and whether the individual has the condition haemophilia

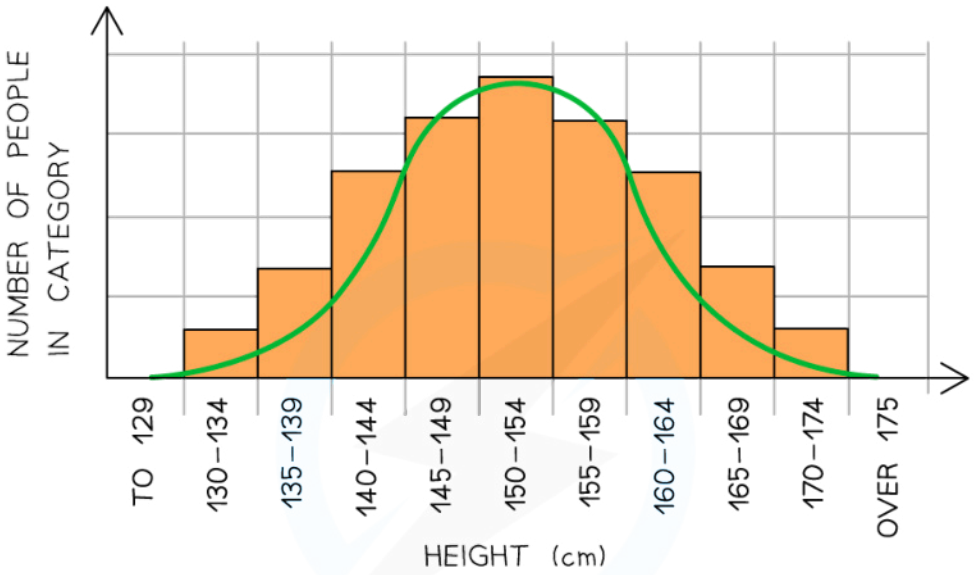

Your notes

Continuous Variation


Your notes

- **Continuous variation** occurs when **two or more genes** affect the final characteristic
- For example, height in humans is determined by **many genetic factors**:
  - Bone length
  - Skeletal muscle structure
  - Ability to absorb food substances effectively
  - Hormone production
  - ...As well as environmental factors like diet, exercise, prenatal nutrition, lifestyle etc
- Most characteristics are determined by more than one gene - a **polygenic** characteristic
- Even **grouped data** like shoe size appears to be discrete but in fact, peoples' feet vary continuously in size
  - Shoe size is merely a practicality for shoe manufacturers, who cannot make exactly the right-sized shoes for everybody
- Continuous variation in birth mass results in the population displaying a **normal distribution** (bell-shaped curve)
  - Of course, environmental factors can affect birth mass, eg. mother's diet, presence of a twin, smoking etc
- Continuous variation occurs when there are **quantitative differences** in the phenotypes of individuals within a population for particular characteristics
- Quantitative differences do not fall into discrete categories like in discontinuous variation
-
  - For example, the mass or height of a human is an example of continuous variation
  - Instead for these features, a **range of values** exist between two extremes within which the phenotype will fall
- The lack of categories and the presence of a range of values can be used to identify continuous variation when it is presented in a table or graph

Your notes



FEATURES OF CONTINUOUS VARIATION:

- NO DISTINCT CLASSES OR CATEGORIES EXIST
- CHARACTERISTICS CAN BE MEASURED AND FALL WITHIN A RANGE BETWEEN TWO EXTREMES

Graph showing population variation in height: an example of continuous variation with quantitative differences

Genetic basis of continuous variation

- This type of variation is caused by an **interaction between genetics and the environment**
- Phenotype = **genotype + environment**
- At the genetic level:
  - Different **alleles** at a single locus have a **small effect** on the phenotype
  - Different **genes** can have the **same effect** on the phenotype and these add together to have an **additive effect**
  - If a large number of genes have a combined effect on the phenotype they are known as **polygenes**

Comparison of Continuous and Discontinuous Variation Table

Feature	Continuous variation	Discontinuous variation
Definition	Features can be measured across a complete range (from one extreme to another). Data collected are quantitative data	Features form distinct classes or categories. Data collected are qualitative data (i.e. discrete or categorical data)
Gene locus	Many loci (that may be on different chromosomes)	Usually only one but may be a very small number
Number of alleles	Many pairs of alleles as many genes contribute to the inheritance (polygenic)	Usually only one pair of alleles (monogenic) but may be a very small number
Effect on phenotype	Many intermediate phenotypes between the extremes (e.g. between shortest and tallest)	Feature either present or absent (the differences are discrete categories)
Environmental influence	Environment has a significant influence	Environment has little to no influence
Examples	Height in humans, milk yield in cattle	Ability to roll tongue, human blood groups


Your notes

Environmental Influence & Variation

Polygenic traits such as human height may also be influenced by environmental factors

- Many environmental factors can affect the intraspecific variation displayed by an organism, including
  - Diet
  - Lifestyle
  - Exercise
  - Exposure to sunlight eg. tanned skin
  - Availability of soil minerals in plants
  - Human intervention eg. pruning plants, neutering animals
  - Fashion, individual preference
  - Native language and dialect (based on where an individual is brought up)
- These traits and differences have been observed in **identical twins** who were unfortunate enough to have been **separated at birth**
  - Not a practice condoned in the 21st century, but was once considered a valid investigative method
- Individuals displayed **distinct phenotypic differences** based on their diet and lifestyle differences


Your notes