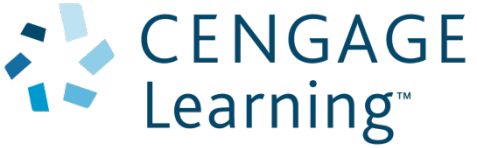
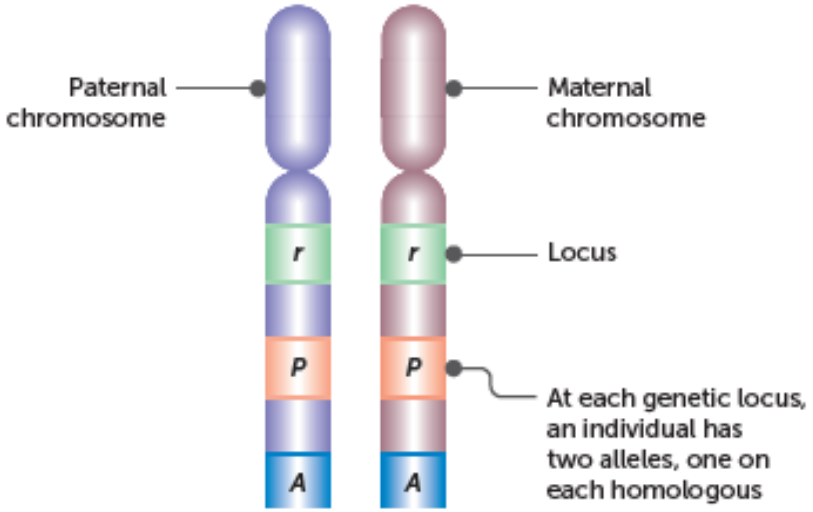
Chapter 5: Genetics



Genetics terminology

Gene
the stored set of instructions for a protein, found on a specific locus (position) on a chromosome



Allele
A form of a gene; different alleles code for the same trait but different versions of the trait. Pairs of alleles are found on a set of maternal and paternal homologous chromosomes.

Genotype
The genetic composition of an organism for a particular trait; the set of alleles (one from each parent) that an organism has for a particular trait

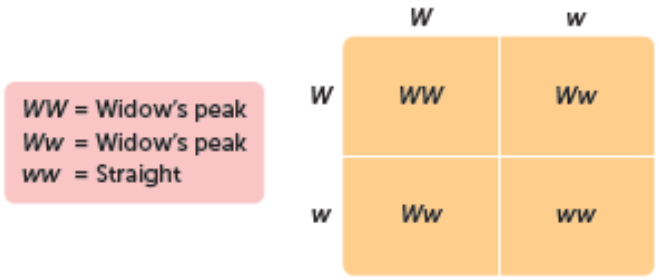
Genetics terminology

Dominant allele

- Always expressed in the phenotype
- Masks a recessive allele if paired with one
- Has the same effect on the phenotype whether it is paired with the same allele or a different one
- Represented by a capital letter

Recessive allele

- Only expressed in the phenotype when present with the same allele (homozygous); e.g. *ww*
- Is masked by a dominant allele
- Represented by a lower-case letter



Genetics terminology

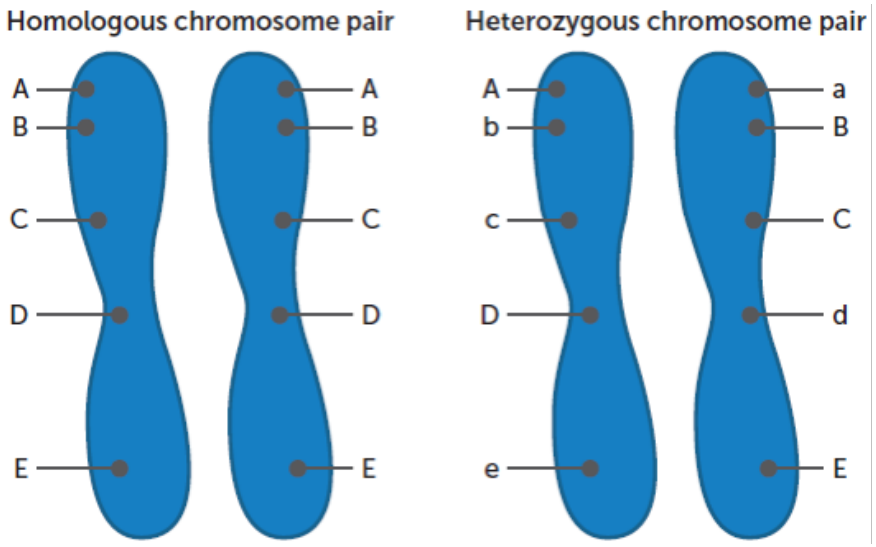
Homozygous
Possessing two identical alleles of a gene

AA is a homozygous dominant genotype

aa is a homozygous recessive genotype

Heterozygous
Possessing two different alleles of a gene; dominant allele will be expressed

Aa is a heterozygous genotype



Pure breed
Identical set of alleles

Genetics terminology

Punnet square

A diagram that shows all possible combinations of alleles and, therefore, all possible genotypes of offspring

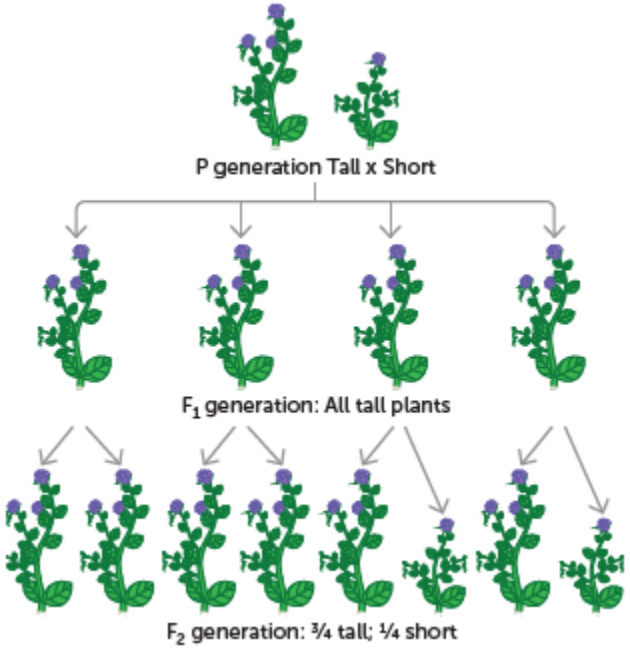
	T	*t*
T	*TT*	*Tt*
t	*Tt*	*tt*

Key:
Alleles
T = tall (dominant allele)
t = short (recessive allele)
Cross
Parent cross = *Tt* × *Tt*

Genotype ratio:	1*TT* : 2*Tt* : 1*tt*	OR	1/4*TT* : 1/2*Tt* : 1/4*tt*	OR	25%*TT* : 50%*Tt* : 25%*tt*
Phenotype ratio:	3 tall : 1 short	OR	¾ tall : ¼ short	OR	75% tall : 25% short

Mendel's experiments

In one of Mendel's experiments, he took a pure-breeding tall pea plant and crossed it with a pure-breeding short pea plant. Pure-breeding plants are ones that, when crossed among themselves, always give rise to offspring that are like the parents. This was one of many experiments conducted by Mendel, leading to the principles of inheritance.



Monohybrid cross: inheritance of a single autosomal gene

A **monohybrid cross** involves fertilisation between two monohybrids (parents with genotypes consisting of one dominant and one recessive allele). Only one gene is investigated.

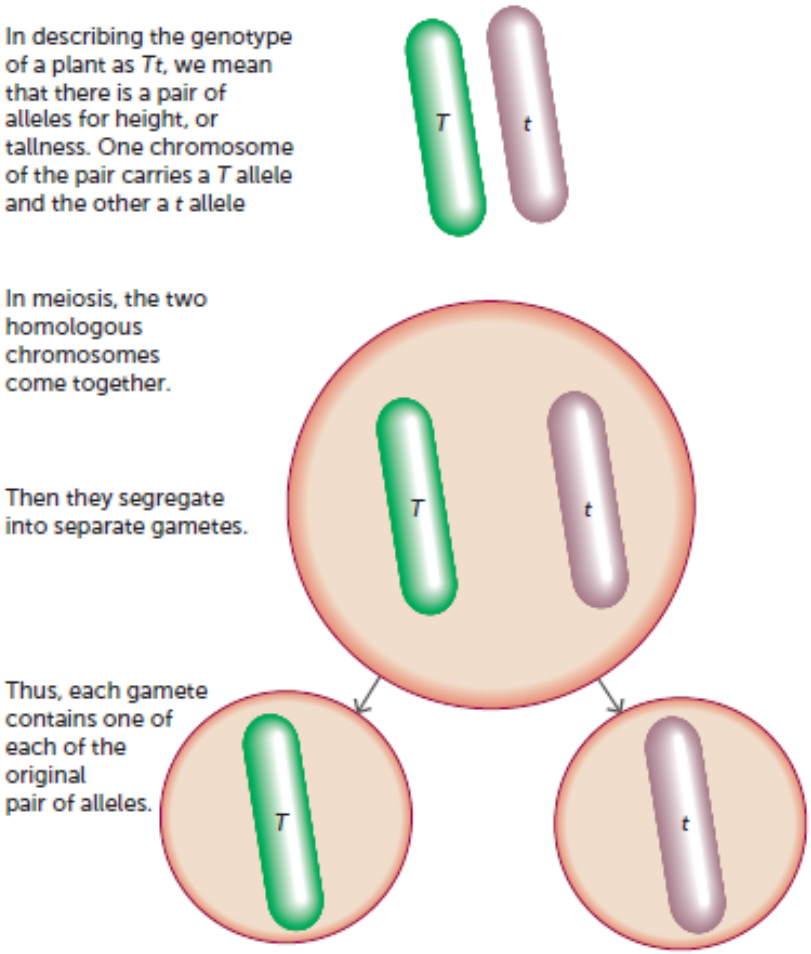
A **monohybrid** is an organism that is heterozygous with respect to a single gene. Monohybrids are the offspring from a cross between parents who are both homozygous but for two different alleles.

Mendel performed crosses with pure-breeding pea plants, which produced monohybrids heterozygous for the gene of interest.

This was the F_1 generation. When he crossed two organisms of the F_1 generation, he was crossing two monohybrids.

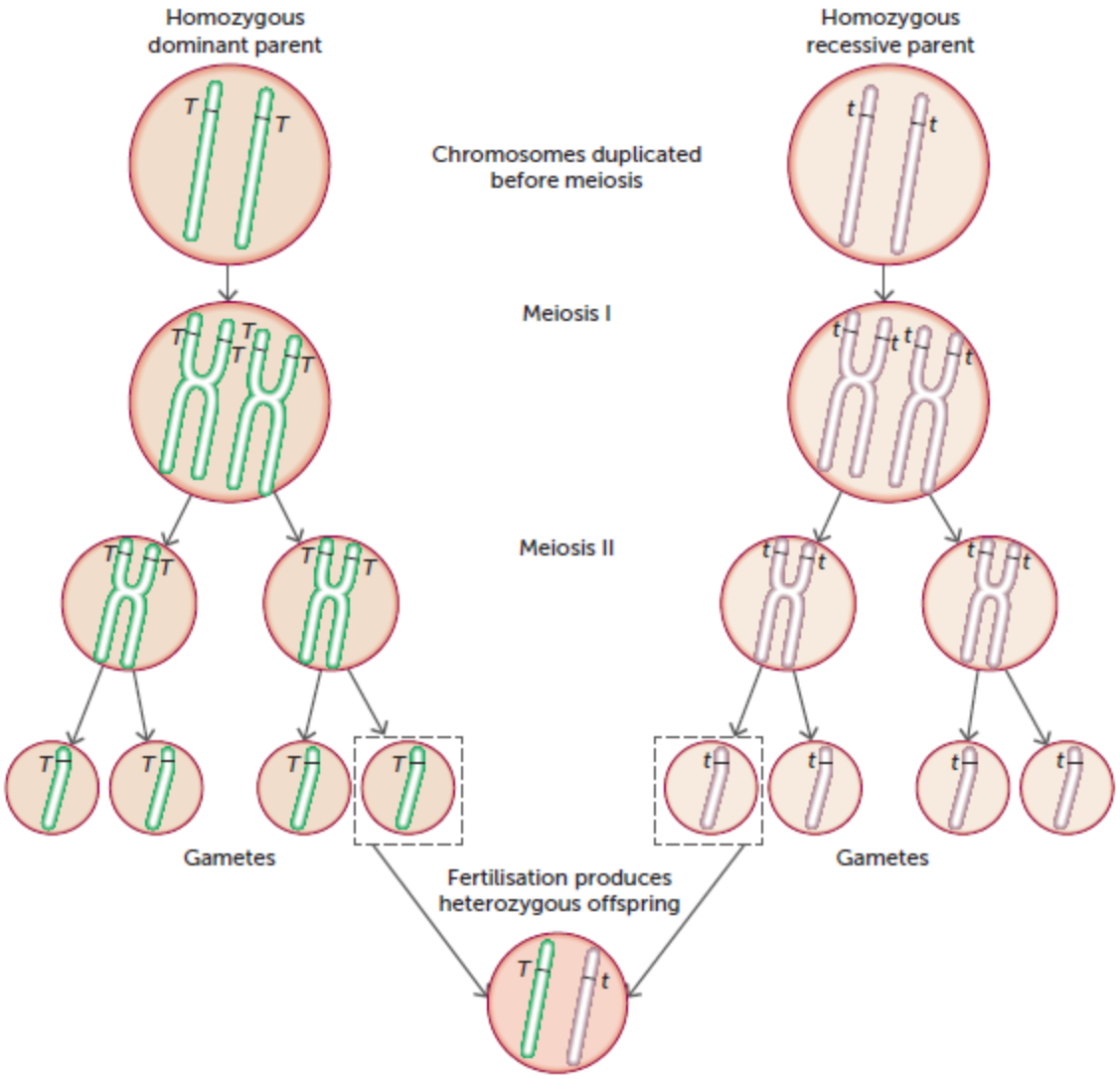
The offspring of the monohybrid cross, known as the F_2 generation, gave rise to a 3:1 ratio of the dominant and recessive phenotypes.

Genetics introduction



The segregation of alleles in inheritance corresponds to the segregation of homologous chromosomes in meiosis.

Genetics introduction



Two homozygous parents with different phenotypes can only produce heterozygous offspring.

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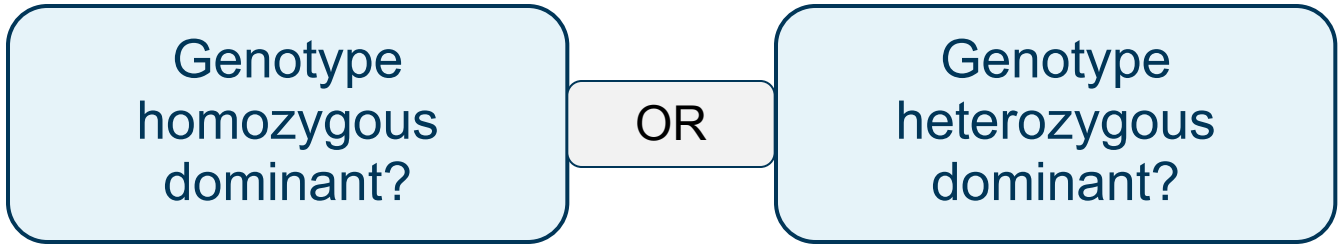
Test cross

If an organism's genotype is unknown, and it is displaying a dominant phenotype, the genotype can be determined by performing a **test cross**.

The cross is usually with an organism that is homozygous recessive at the locus in question.

The ratio of phenotypes in the offspring reveals the unknown genotype.



Determining genotype: example

In guinea pigs, black fur colour is dominant over white. A test cross could be performed to find out the genotype of a black pet guinea pig with an unknown genotype. If the owner wanted to be certain their pet was homozygous dominant black (for breeding purposes), they could breed the pet with a guinea pig who was homozygous recessive white.

Either 100 per cent or 50 per cent of the offspring will present the dominant phenotype. There are two possible crosses, given the black phenotype could have arisen from the homozygous or heterozygous set of alleles.

Determining genotype: example

Black guinea pig's genotype: *BB*

	B	*B*
b	*Bb*	*Bb*
b	*Bb*	*Bb*

Genotype ratio:	100% *Bb*
Phenotype ratio:	100% black

Black guinea pig's genotype: *bb*

	B	*b*
b	*Bb*	*bb*
b	*Bb*	*bb*

Genotype ratio:	50% *Bb* : 50% *bb*
Phenotype ratio:	50% black : 50% white

Multiple alleles in contrast to monohybrid inheritance

For most traits, there are more than two forms of alleles for a gene. This is known as **multiple alleles**.

In any one individual, only two alleles are normally present, a monohybrid. A multiple allele system is present when three or more alleles of a gene exist among the members of a population.

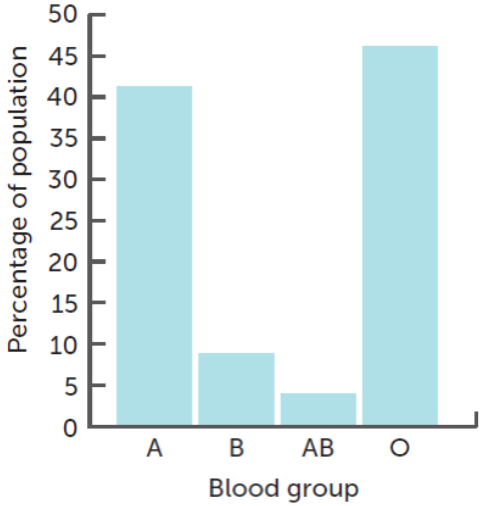
Multiple alleles result in discontinuous variation

An example of this is seen in the ABO blood group system in humans. In the human population, there are three alleles possible for one gene.

There are four possible phenotypes, with no variation in between each blood group. This leads to **discontinuous variation**, because only one set of alleles for one gene determines the phenotype. Discontinuous variation is a set of discrete phenotypic categories controlled by a single gene and its set of alleles.

Phenotypes and genotypes for human blood groups

Phenotype	Genotype
Blood type A	I^AI^A or I^Ai
Blood type B	I^BI^B or I^Bi
Blood type AB	I^AI^B
Blood type O	ii



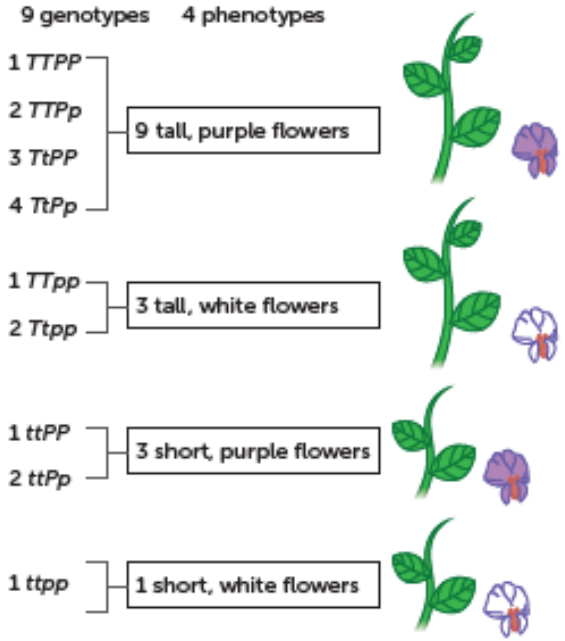
Inheritance of two independently inherited autosomal genes

A **dihybrid cross** involves two genes with two different alleles for each gene.

Inheritance for two unlinked autosomal genes can be analysed with a dihybrid cross. If the P generation are pure-breeding with respect to all traits, the F2 generation typically shows a ratio of 9:3:3:1.

The basic structure of a dihybrid cross Punnett square is a 4×4 grid for possible offspring, with the four possible gamete allele combinations of one parent written across the top row and those of the other parent written down the left column. A key is drawn up for the alleles and the cross.

Dihybrid cross: flower colour and stem length



Possible gametes (same for both parents):

	TP	*Tp*	*tP*	*tp*
TP	*TTPP*	*TTPp*	*TtPP*	*TtPp*
Tp	*TTPp*	*TTpp*	*TtPp*	*Ttpp*
tP	*TtPP*	*TtPp*	*ttPP*	*ttPp*
tp	*TtPp*	*Ttpp*	*ttPp*	*ttpp*

Polygenic inheritance defined

For one characteristic, two or more genes and therefore two or more sets of alleles contribute to a phenotype.

A characteristic controlled by more than one gene is known as a polygenic characteristic, and its transmission is called **polygenic inheritance**.

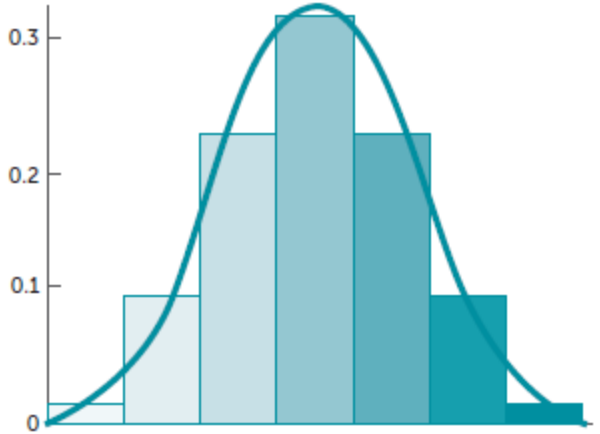
Polygenes consist of multiple genes, and each gene consists of multiple alleles.

Polygenic inheritance

Polygenic inheritance example

An example of polygenic inheritance is human height.

Humans have a range of heights, with a smooth gradation from one extreme to another. This can be seen when you line up for your school photographs each year. The condition of showing a range of phenotypes is called **continuous variation**.



Comparing continuous and discontinuous variation

Factor	Continuous variation	Discontinuous variation
Type of inheritance that may lead to this variation	Single-gene inheritance (but may have multiple alleles)	Polygenic inheritance (will have multiple alleles for each gene and several genes)
Cause	Genetic and environmental factors	Genetic factors
Description	Variation that shows gradual changes from one trait to another. Differences are slight, along a continuum.	Variation that shows clear and discrete changes between traits. There is no intermediate form.
Type of graphical representation	Histogram (a line graph can be superimposed to show continuous data)	Bar graph
Examples	Height, weight, skin colour	Ability or inability to tongue roll; attached or detached ear lobes; blood groups A, B, AB and O

Dominant inheritance patterns

Incomplete dominance

Incomplete dominance occurs when two different alleles are present, but neither allele is completely dominant.

Both alleles contribute to the phenotype, but only partially.

A third intermediary phenotype is observed.

Example:

In pure-breeding snapdragons, incomplete dominance of the red flower colour and white flower colour results in a pink flower colour.

Science Photo Library/Adrian Thomas

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Dominant inheritance patterns

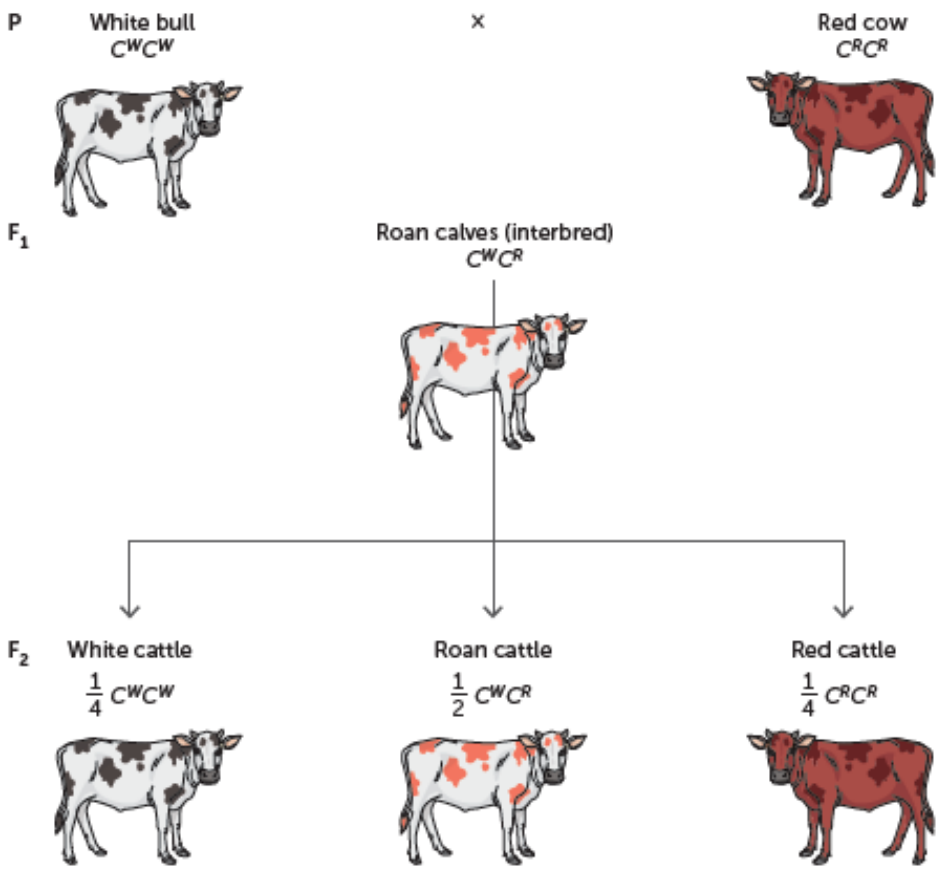
Codominance

Codominance occurs when two alleles are completely dominant.

Both alleles are observed and expressed in the phenotype.

Example:

In shorthorn cattle, codominant inheritance results in a roan coat colour in the offspring of pure-breeding red and white parents.



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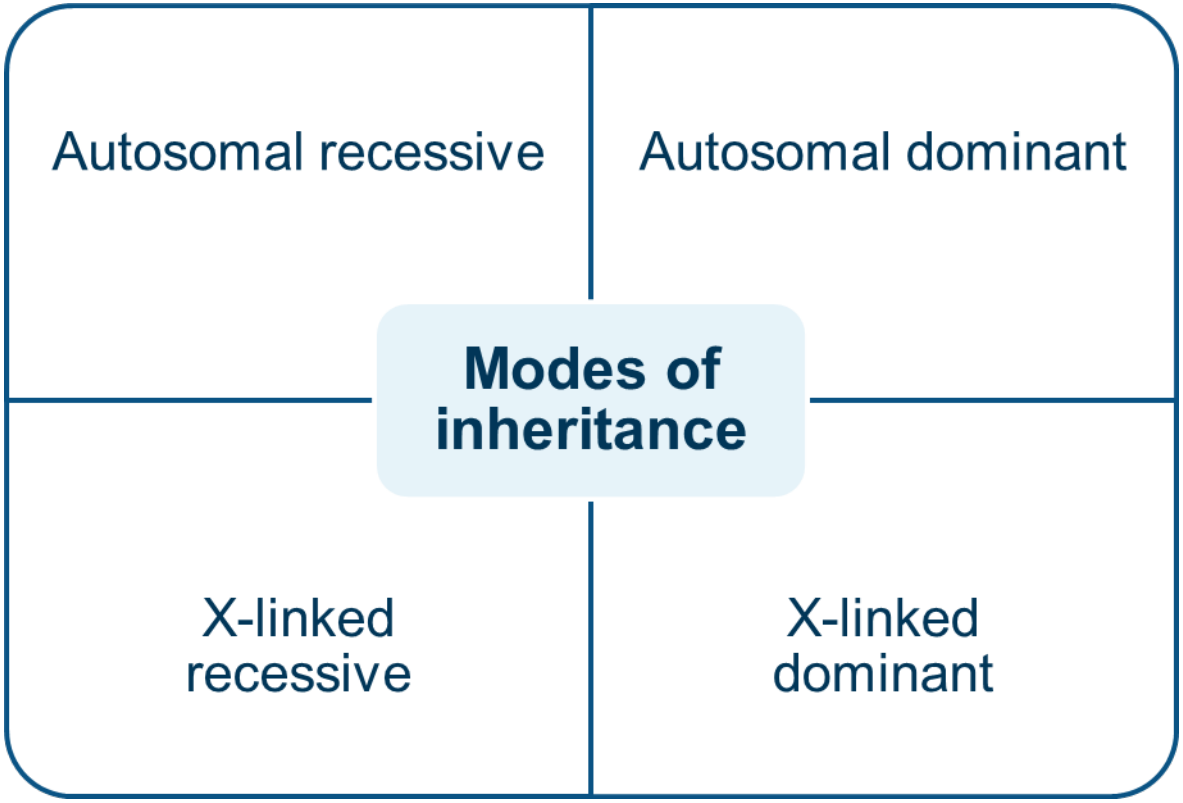
Dominant inheritance patterns

Symbolism conventions for complete, incomplete and codominance

	Complete dominance	Incomplete (partial) dominance	Codominance
Parents	*BB, bb*	C^BC^B, C^WC^W	C^BC^B, C^WC^W
Gametes	*Bb*	C^BC^w	C^BC^w
F1 genotype	*Bb*	C^BC^W	C^BC^W
Phenotype	Black	Grey	Black and white patches
Gametes	*Bb, Bb*	C^BC^W, C^BC^W	C^BC^W, C^BC^W
F2 genotype	*BB, Bb, bb*	C^BC^B, C^BC^W, C^WC^W	C^BC^B, C^BC^W, C^WC^W
Phenotype	Black White	Black Grey White	Black Black and white patches White
Heterozygote	Same as dominant	Intermediate between homozygous parents	Properties of both homozygous parents

Note: B and C^B = alleles for black coat colour; *b* and C^W = alleles for white coat colour

Four main modes of inheritance



Autosomal recessive patterns

An allele on a non-sex chromosome being passed to offspring is known as **autosomal inheritance**.

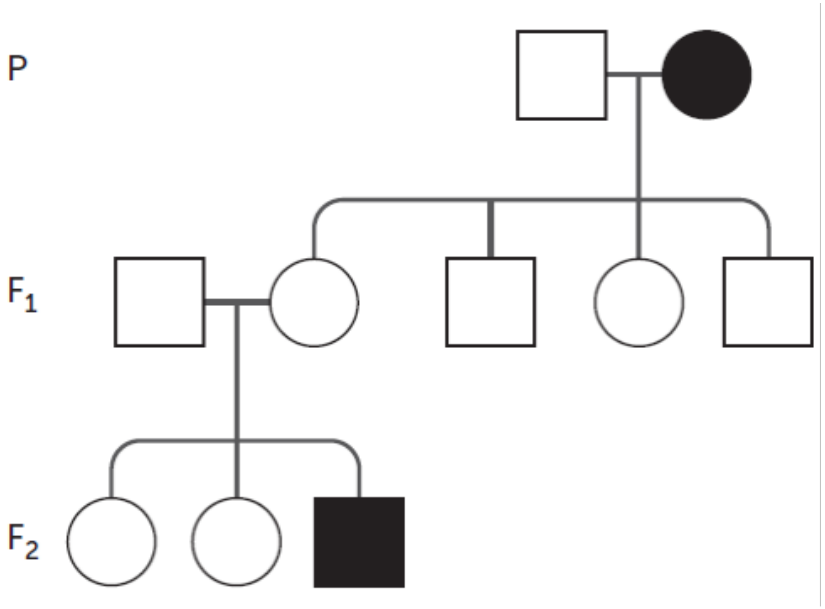
People with only one defective allele in the pair of alleles are called carriers.

These people are most often not affected with the condition. They can pass the abnormal gene on to their children.

The parents of an affected person are always at least carriers of the allele.

A carrier is usually unaffected, because a dominant allele will silence the effects of the recessive allele that causes the condition.

Autosomal recessive patterns: example



Two unaffected parents in the F_1 generation had an affected son. At least one of the parents would need to have had the allele, causing the condition to pass on to the son. If the allele was dominant, the F_1 parent would have been affected. As the parents are carriers, rather than affected by the condition, the allele must be recessive.

Autosomal dominant patterns

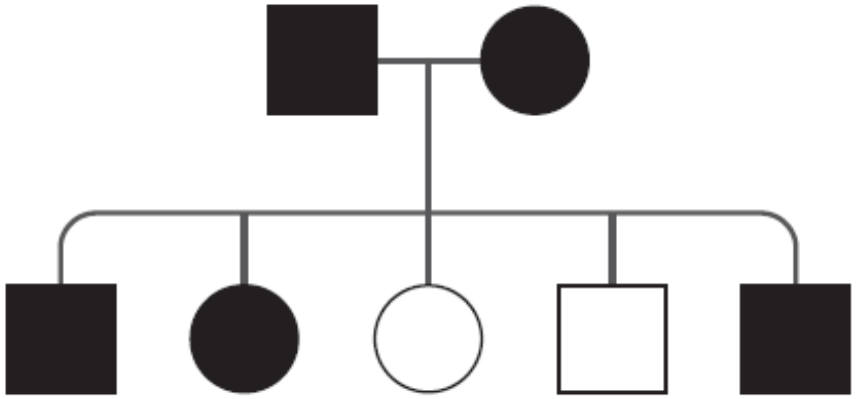
In autosomal dominant inheritance, a single dominant allele is responsible for the occurrence of a phenotype.

Each affected person usually has an affected parent, and the phenotype occurs in every generation.

A single copy of the affected allele is enough to cause the condition. A parent with a single copy of a dominant allele on an autosome (heterozygous) will theoretically pass it on to 50% of the offspring.

If the parent is homozygous for the dominant trait, then 100% of the offspring will be affected.

Autosomal dominant patterns: example



The trait affecting individuals is a dominant trait. We know it is dominant because two of the offspring are unaffected, yet both parents are affected. This means both parents were able to pass a recessive ‘normal’ allele to the unaffected offspring, which means both parents are heterozygous and affected.

X-linked recessive patterns

When a recessive phenotype under investigation is determined by an allele on the X chromosome, it is said to be an **X-linked** recessive phenotype.

Males who have the recessive allele on their X chromosome will always express the phenotype, because they only have one X chromosome.

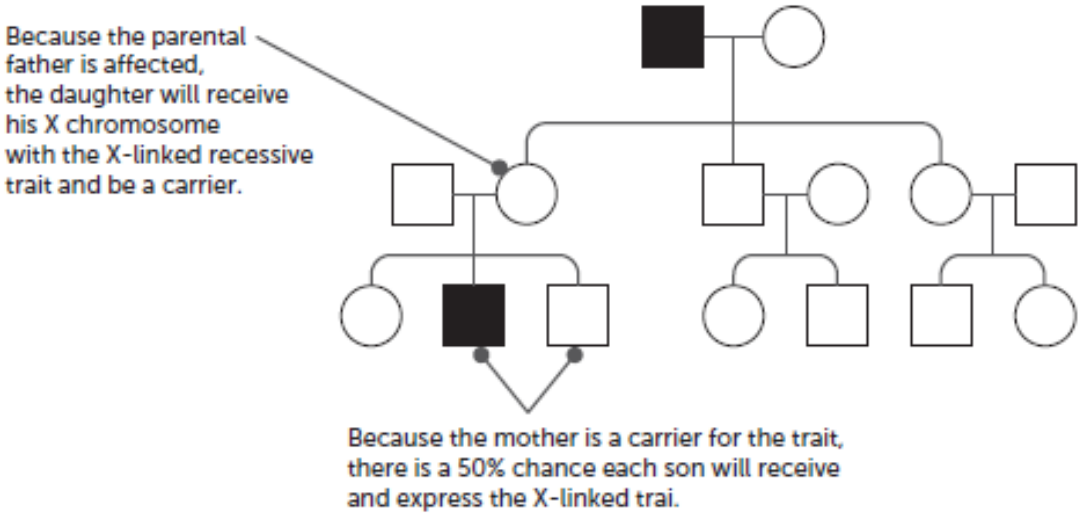
Females will only express the phenotype when both X chromosomes have the affected allele.

For a female child to be affected, the father must be affected and the mother must be either affected or a carrier.

A heterozygous female will be a carrier.

Modes of inheritance

X-linked recessive patterns: example



Source: Genetics Generation

X-linked dominant patterns

The heterozygous females will always show the phenotype, and any individuals with the phenotype must have a parent with the phenotype.

Males showing the phenotype will not pass the affected allele on to their sons (because they must inherit their father's Y chromosome), but they will pass it on to all their daughters, who will also show the phenotype. This is because daughters always inherit their father's X chromosome.

A heterozygous female is expected to pass on the allele to 50% of her offspring, regardless of their sex.

The condition should appear in every generation.

An affected male receives the dominant allele from an affected mother.

Y-linked patterns

If a trait is carried on the Y chromosome, it is said to be Y-linked.

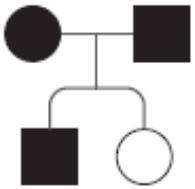
Only males are affected.

The most conspicuous phenotype associated with genes on the Y chromosome is male gender.

Modes of inheritance

Inheritance patterns determined in a pedigree

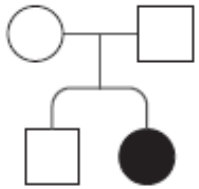
AUTOSOMAL DOMINANT



The allele cannot be recessive as two affected parents could **not** have an unaffected offspring.

The parents MUST be heterozygous

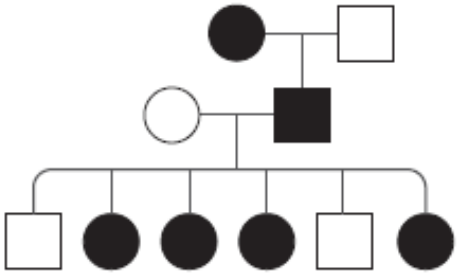
AUTOSOMAL RECESSIVE



The allele cannot be dominant as two unaffected parents could **not** have an affected offspring.

The parents MUST be heterozygous

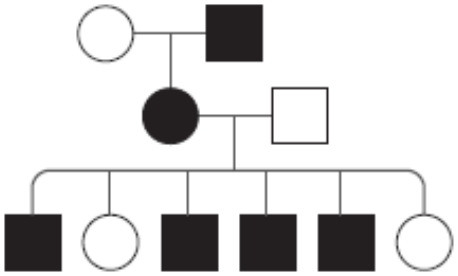
X-LINKED DOMINANT



Sex linkage cannot be confirmed.

100% incidence of affected daughters from an affected father *suggests* X-linked dominance.

X-LINKED RECESSIVE



Sex linkage cannot be confirmed.

100% incidence of affected sons from an affected mother *suggests* X-linked recessive.

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Mode of inheritance in a pedigree using a dichotomous key