

Heredity and Genetics

Matthew Williams • Biology • May 9, 2026

Heredity and Genetics

Genetics is the study of how characteristics are transmitted from parent to offspring. Traits are controlled by **genes** — units of inheritance located at specific positions (loci) on chromosomes. Each gene may exist in alternative forms called **alleles**, and the combination an individual inherits determines their genetic makeup.

Core Terminology

These terms appear throughout genetics problems and must be defined precisely.

Term	Definition
Gene	A unit of inheritance controlling a particular characteristic
Allele	An alternative form of a gene
Genotype	The genetic composition for a trait (e.g. Tt, TT, tt)
Phenotype	The observable expression of the genotype
Dominant allele	Expressed in the phenotype when one or two copies are present
Recessive allele	Expressed only when two copies are present (no dominant allele)
Homozygous	Two identical alleles (e.g. TT or tt)
Heterozygous	Two different alleles (e.g. Tt)

Mendelian Inheritance

Gregor Mendel established the principles of inheritance through controlled crosses with pea plants. His key insight was that traits are governed by discrete factors — alleles — that separate during reproduction and recombine randomly.

Mendel's F1 and F2 pea plant crosses

When a pure-breeding tall plant (TT) was crossed with a pure-breeding dwarf plant (tt), all F1 offspring were tall — the tallness allele is dominant. When F1 plants (Tt) were crossed with each other, the F2 generation showed a 3:1 ratio of tall to dwarf. This result is a cornerstone of monohybrid inheritance.

Law of Segregation: Allele pairs separate during gamete formation so each gamete carries only one allele. Alleles recombine randomly at fertilisation.

Punnett Squares

A Punnett square maps all possible allele combinations from a cross. Each parent's gametes line the top and side; the boxes show possible offspring genotypes.

Punnett square for Tt × Tt cross

For Tt × Tt:

	T	t
T	TT	Tt
t	Tt	tt

- Genotype ratio: 1 TT : 2 Tt : 1 tt
- Phenotype ratio: 3 tall : 1 dwarf

These ratios express probability, not guaranteed outcomes. Small sample sizes and unequal survival can shift observed ratios away from the theoretical values.

Types of Dominance

Not all alleles follow simple dominant-recessive relationships.

Type	Description	Example	F1 phenotype	F2 ratio
Complete dominance	One allele fully masks the other	Pea plant height	Dominant only	3:1
Incomplete dominance	Heterozygote is intermediate	Snapdragon colour	Blend (e.g. pink)	1:2:1
Codominance	Both alleles fully expressed together	ABO blood groups (I ^A I ^B)	Both traits visible	1:2:1

Incomplete dominance — snapdragon: Red (RR) × White (rr) 'all Pink (Rr) in F1. Crossing two pink plants: 1 Red : 2 Pink : 1 White.

Codominance — Shorthorn cattle: Red × White 'Roan (cattle with both red and white hairs), because both alleles are expressed simultaneously.

Multiple Alleles: ABO Blood Groups

The ABO system is controlled by three alleles — I^A , I^B , and I^O — where I^A and I^B are codominant and I^O is recessive to both.

Blood Group	Possible Genotypes	Antigens on RBC
A	$I^A I^A$ or $I^A I^O$	A antigen
B	$I^B I^B$ or $I^B I^O$	B antigen
AB	$I^A I^B$	A and B antigens
O	$I^O I^O$	None

Blood group O is the universal donor (no antigens to trigger rejection); AB is the universal recipient (neither A nor B antibodies in plasma).

Test Cross

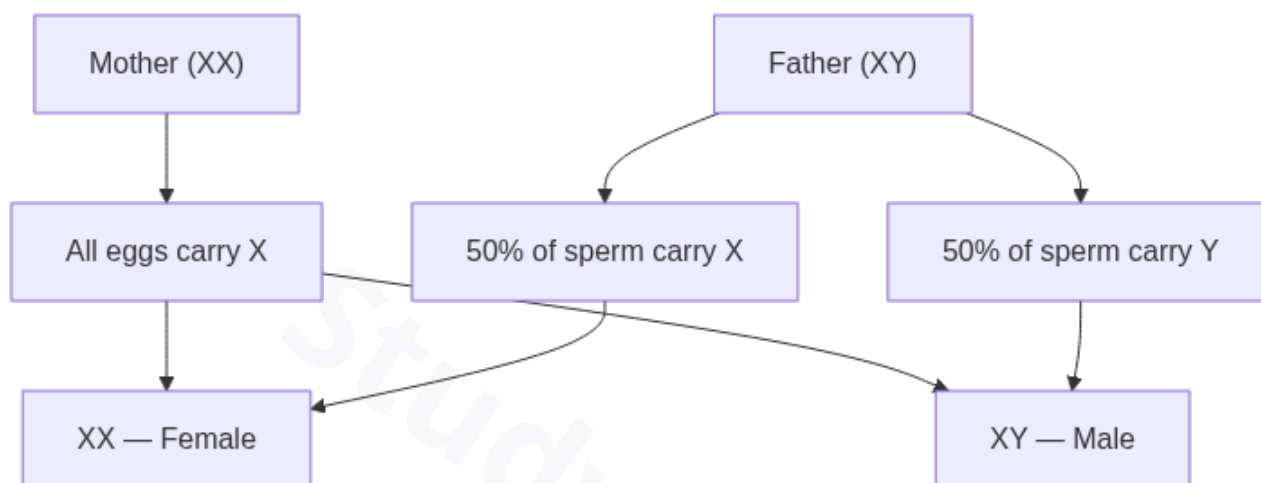
A test cross determines the genotype of an organism showing the dominant phenotype by crossing it with a homozygous recessive individual.

Test cross outcomes

- All offspring show dominant phenotype 'unknown parent is **homozygous dominant** (AA)
- 1:1 ratio of dominant to recessive offspring 'unknown parent is **heterozygous** (Aa)

Sex Determination

In humans, sex is determined by the sex chromosomes. Females carry two X chromosomes (XX); males carry one X and one Y (XY). Eggs always carry X; sperm carry either X or Y — so it is the sperm that determines the sex of the offspring.



Sex determination in humans

The probability of producing a male or female offspring is equal (approximately 1:1) in any given pregnancy.

Sex-Linked Inheritance

Genes located on the X chromosome are described as **sex-linked**. Because males have only one X chromosome, they express any allele on it — including recessive ones — without needing a second copy. This is why sex-linked recessive conditions are far more common in males.

Genotype	Individual	Status
$X^{H}X^{H}$	Female	Unaffected
$X^{H}X^{h}$	Female	Carrier (unaffected, but carries recessive allele)
$X^{h}X^{h}$	Female	Affected
$X^{H}Y$	Male	Unaffected
$X^{h}Y$	Male	Affected

Colour blindness (red-green) is the most common sex-linked recessive condition examined at CSEC. The normal vision allele (X^H) is dominant over the colour-blind allele (X^h).

Example cross: carrier female \times unaffected male ($X^H X^h \times X^H Y$)

	X^H	Y
X^H	$X^H X^H$ (unaffected female)	$X^H Y$ (unaffected male)
X^h	$X^H X^h$ (carrier female)	$X^h Y$ (colour-blind male)

Result: daughters are unaffected (though one in two is a carrier); 50% of sons are colour-blind.

Haemophilia follows the same pattern — a sex-linked recessive condition in which blood fails to clot normally.

Exam Tip

Always write sex-linked genotypes with the X and Y chromosomes explicit — X^H X^h , not just Hh. This makes clear to the examiner that you understand the gene is carried on the X chromosome. Showing the Punnett square with X and Y columns and rows earns full method marks even if one offspring genotype is wrong.

Pedigree Analysis

A pedigree chart traces the inheritance of a trait through multiple generations. Circles represent females; squares represent males; shaded symbols indicate affected individuals; horizontal lines connect parents; vertical lines lead to offspring.

Pedigree chart showing an autosomal recessive inheritance pattern

Observed pattern	Interpretation
Two unaffected parents ' affected child	Trait is recessive (both parents are carriers)
Trait appears in every generation	Trait is likely dominant
Trait skips generations	Trait is recessive
Affected individuals mostly male	Possibly sex-linked recessive
Father-to-son transmission	Cannot be X-linked (fathers pass Y to sons)

When interpreting pedigrees, start by determining whether the trait is dominant or recessive, then decide whether it is autosomal or sex-linked.

Summary of Key Principles

- Traits are controlled by alleles on chromosomes; dominant alleles are expressed over recessive ones
- Mendelian ratios (3:1, 1:2:1, 1:1) describe probability of offspring phenotypes, not guaranteed outcomes
- Incomplete dominance and codominance modify the standard 3:1 ratio to 1:2:1
- Sex is determined by sex chromosomes; sex-linked genes are on the X chromosome and show different patterns in males and females
- Pedigree charts allow inference of genotype and inheritance pattern across generations

PRACTICE — GENETICS CORE TERMS

Gene

A unit of inheritance controlling a particular characteristic, located at a specific locus on a chromosome.

Allele

An alternative form of a gene.

Genotype

The genetic makeup of an organism for a trait (e.g. Tt).

Phenotype

The observable expression of the genotype, shaped by the environment.

Homozygous

Having two identical alleles for a trait (e.g. TT or tt).

Heterozygous

Having two different alleles for a trait (e.g. Tt).

Dominant allele

An allele expressed in the phenotype whenever one copy is present.

Recessive allele

An allele expressed only when two copies are present and no dominant allele exists.

Incomplete dominance

A pattern where the heterozygote phenotype is intermediate between both homozygous forms.

Codominance

A pattern where both alleles are fully expressed simultaneously in the heterozygote.

Test cross

A cross between an organism of unknown genotype (dominant phenotype) and a homozygous recessive individual to determine the unknown genotype.

Sex-linked gene

A gene carried on the X chromosome; males express recessive sex-linked alleles because they have only one X.

Carrier

A heterozygous individual who carries a recessive allele but does not express the associated condition.

Pedigree chart

A diagram tracing the inheritance of a trait across multiple generations of a family.

Law of Segregation

Mendel's principle that allele pairs separate during gamete formation so each gamete carries only one allele.