Collegedunia NCERT Notes

The Ultimate NCERT Revision Guide for Class 12 Biology

Chapter 4: Principles of Inheritance and Variation

NCERT Class 12 Biology — New NCERT (2026-27) — Full-colour diagrams

Also see for this chapter: **NCERT Solutions** | **Formula Sheet** | **Exemplar Solutions**

How to use these notes: Genetics is one of the highest-yielding units for both the CBSE board and NEET. This chapter alone contributes a steady block of questions every year — Mendelian ratios, pedigree analysis, blood-group genetics and chromosomal disorders are perennial favourites. Work through each section in order; the worked Punnett squares and the disorder comparison tables are designed to be revised the night before the exam.

Contents

1 Foundations of Genetics and Mendel

Genetics is the branch of biology that studies **inheritance** (the transmission of characters from parents to offspring) and **variation** (the differences progeny show from their parents and from each other). Heredity explains why an elephant gives birth only to an elephant; variation explains why siblings are not identical. Long before any scientific basis was known — from roughly 8000–1000 B.C. — humans had already exploited natural variation through artificial selection and domestication, producing breeds such as the Sahiwal cow of Punjab.

The systematic study began with **Gregor Johann Mendel**, who carried out hybridisation experiments on the garden pea (*Pisum sativum*) for seven years (1856–1863). Mendel was the first to apply statistical analysis and mathematical logic to a biological problem. His large sample sizes and his confirmation of results across successive generations gave his conclusions the weight of general rules rather than isolated observations.

1.1 Why the garden pea was the perfect choice

Mendel's choice of organism was as important as his method. The pea plant offered several decisive advantages.

- Many **contrasting traits** with clear, distinct alternatives (no blending), e.g. tall vs dwarf, round vs wrinkled seed.
- **Naturally self-pollinating**, so true-breeding lines are easy to maintain; the flower structure also permits controlled artificial cross-pollination.
- **Short life cycle** and large number of offspring per cross, giving statistically reliable ratios.
- Easy to grow and maintain; results could be reproduced over many generations.

A **true-breeding line** is one that, after continuous self-pollination, shows stable inheritance of a trait for several generations. Mendel selected 14 true-breeding varieties forming 7 pairs, each pair differing in exactly one character with two contrasting traits.

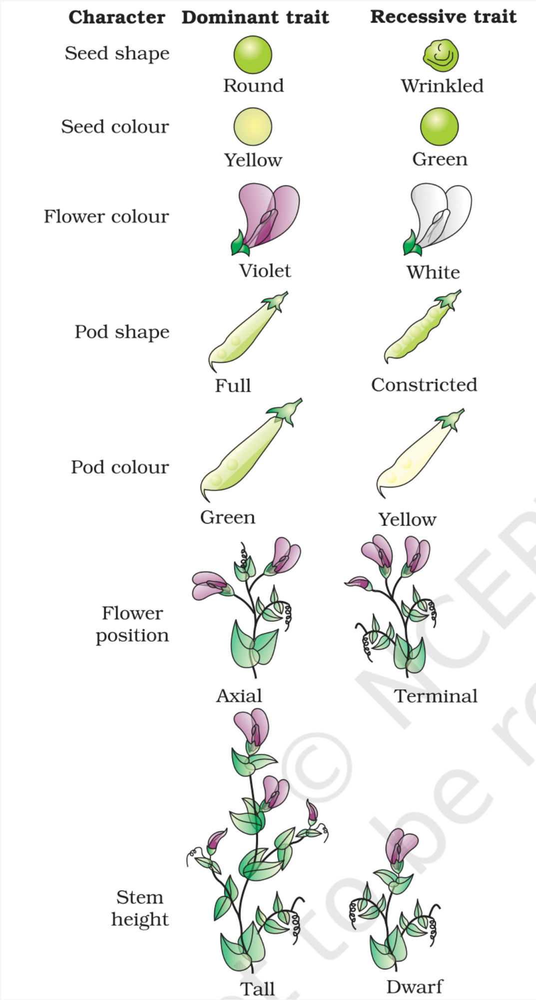


Fig 4.1 — The seven pairs of contrasting traits in the garden pea studied by Mendel, showing the dominant and recessive form of each character.

The Seven Characters Mendel Studied

Stem height (tall/dwarf), flower colour (violet/white), flower position (axial/terminal), pod shape (inflated/constricted), pod colour (green/yellow), seed shape (round/wrinkled), and seed colour (yellow/green). Each character had two sharply distinct, non-blending traits — this discreteness is what allowed clean numerical ratios.

1.2 Key terminology of classical genetics

These terms recur throughout the chapter and throughout NEET genetics questions. Fix them firmly before moving on.

Term	Meaning
Gene	The unit of inheritance; carries the information to express a trait.
Allele	One of the alternative forms of a gene that codes for a pair of contrasting traits.
Genotype	The genetic (allelic) constitution of an organism, e.g. TT, Tt, tt.
Phenotype	The observable, expressed character, e.g. tall or dwarf.
Homozygous	Both alleles identical (TT or tt); produces one kind of gamete.
Heterozygous	Two different alleles (Tt); produces two kinds of gamete in equal proportion.
Dominant	The allele/trait expressed in the heterozygote (written as a capital letter).
Recessive	The allele/trait masked in the heterozygote, expressed only when homozygous (lower-case letter).

Genotype vs Phenotype

A common slip is to treat "3:1" and "1:2:1" as the same thing. In a monohybrid F_2 the **phenotypic ratio is 3:1** but the **genotypic ratio is 1:2:1**. Always state which ratio the question asks for — examiners deliberately mix the two.

Capital = Dominant

Always use the *same letter* in two cases for an allele pair — T and t, never T and d. The capital is the dominant trait expressed in F_1; the lower-case is recessive. Mixing letters (like T for tall, d for dwarf) makes it impossible to tell whether the two are alleles of the same gene.

2 Inheritance of One Gene — The Monohybrid Cross

When Mendel crossed a true-breeding tall pea (TT) with a true-breeding dwarf pea (tt), every plant in the first filial (F_1) generation was **tall** — none were dwarf, and none were of intermediate height. The dwarf character had completely disappeared. He observed the same pattern for all seven trait-pairs: only one parental trait appeared in F_1.

When the tall F_1 plants were self-pollinated, the dwarf character **reappeared** in the F_2 generation. The ratio was striking and constant: $\frac{3}{4}$ of F_2 plants were tall and $\frac{1}{4}$ were dwarf — a clean **3:1 phenotypic ratio** — with no blended intermediates.

2.1 The Punnett square and the underlying logic

To explain these numbers, Mendel proposed that a heritable “factor” (now **gene**) is passed unchanged through gametes, and that it occurs in pairs. During gamete formation by meiosis, the two alleles **segregate** so that each gamete carries only one allele. The British geneticist Reginald C. Punnett devised the **Punnett square**, a grid for computing all possible offspring genotypes and their probabilities.

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

$F_1 \times F_1$ ($Tt \times Tt$) self-cross

The grid shows that out of four equally likely fertilisations, 1 is TT, 2 are Tt, and 1 is tt. Because T is dominant, TT and Tt both look tall, so phenotypically 3 tall : 1 dwarf, while genotypically $1\,TT : 2\,Tt : 1\,tt$.

Monohybrid Ratios

F_2 phenotypic ratio $= 3 : 1$ F_2 genotypic ratio $= 1 : 2 : 1$

The $\frac{1}{4} : \frac{1}{2} : \frac{1}{4}$ split is the binomial expansion of a pair of gametes carrying T or t at equal frequency $\frac{1}{2}$:

$$\left(\tfrac{1}{2}T + \tfrac{1}{2}t\right)^2 = \tfrac{1}{4}\,TT + \tfrac{1}{2}\,Tt + \tfrac{1}{4}\,tt$$

The complete Mendelian monohybrid cross — showing both the $P \to F_1$ and the F_1 self-cross stages with gametes and ratios — is shown below as drawn in the NCERT textbook.

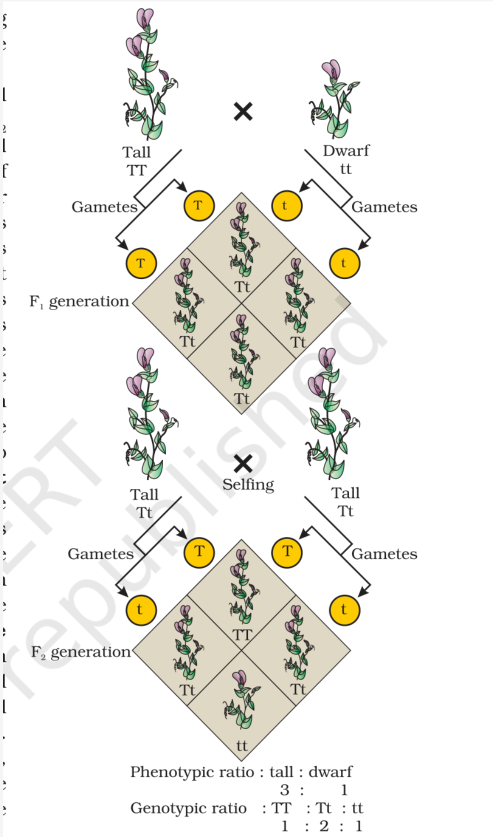


Fig 4.4 — A Punnett square used to understand a typical monohybrid cross between true-breeding tall and true-breeding dwarf plants, with the resulting 3:1 phenotypic and 1:2:1 genotypic ratios.

Segregation is a Meiotic Event

Each parent has two alleles, but during meiosis the allele pair **separates** so that a gamete carries only one. A homozygote (TT or tt) makes one type of gamete; a heterozygote (Tt) makes two types (T and t) in a 1:1 ratio. Random fertilisation then recombines them.

2.2 The test cross

By looking at a tall plant you cannot tell whether its genotype is TT or Tt — the phenotype is identical. To determine the unknown genotype of an organism showing a dominant phenotype, it is crossed with the **homozygous recessive** parent. This is a **test cross**.

Case 1: Tested plant is TT

$TT \times tt$

↓

All offspring Tt
100% tall

No recessive offspring ⇒ parent was homozygous

Case 2: Tested plant is Tt

$Tt \times tt$

↓

$1\,Tt : 1\,tt$
1 tall : 1 dwarf

Recessive offspring appear ⇒ parent was heterozygous

Quick Tip

In a test cross, count the offspring. **All dominant** ⇒ the tested parent is homozygous (TT). **A 1:1 split** of dominant to recessive ⇒ the parent is heterozygous (Tt). This single observation pins down the genotype — no self-cross needed.

2.3 Mendel's First Two Laws

From the monohybrid data Mendel framed two general principles.

Law of Dominance. (i) Characters are controlled by discrete units called factors. (ii) Factors occur in pairs. (iii) In a dissimilar pair, one factor (dominant) masks the other (recessive). This law explains why only one trait shows in F_1 and why the F_2 ratio is 3:1.

Law of Segregation. The two alleles of a pair do not blend; they segregate during gamete formation so that each gamete receives only one allele. Both characters are recovered intact in F_2. A homozygous parent forms only one kind of gamete; a heterozygous one forms two kinds in equal proportion. This law has *no exception* and is therefore also called the Law of Purity of Gametes.

Why Dominance Happens — the Molecular View

Consider a gene coding for an enzyme. A modified (mutant) allele may produce (i) a normal/less efficient enzyme, (ii) a non-functional enzyme, or (iii) no enzyme. If the unmodified allele alone makes enough functional enzyme to give the normal phenotype, that allele is **dominant** and the modified one is **recessive**. Dominance is therefore a property of the gene product, not an inherent label of the gene.

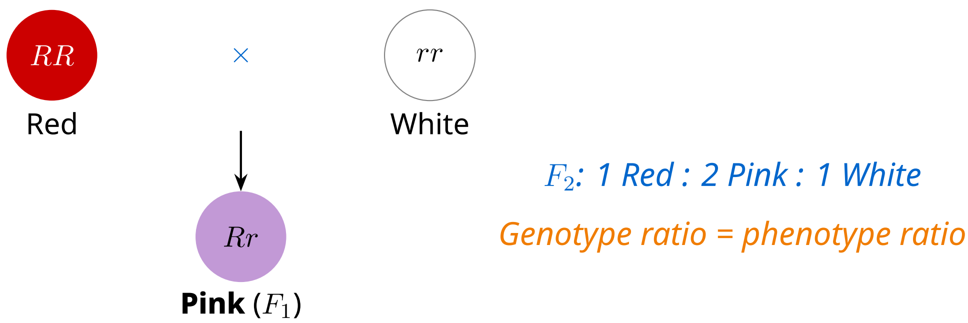
Solve the NCERT Exercises for this Chapter

3 Deviations from Mendelian Dominance

Not every gene behaves with simple complete dominance. Three important deviations — incomplete dominance, co-dominance and pleiotropy — are heavily tested and must be distinguished carefully.

3.1 Incomplete dominance

In the dog flower (snapdragon, *Antirrhinum* sp.), a cross between true-breeding red (RR) and true-breeding white (rr) flowers gives an F_1 that is **pink** (Rr) — intermediate between the two parents, resembling neither. On selfing, F_2 segregates as 1 red (RR) : 2 pink (Rr) : 1 white (rr).



The genotypic ratio is exactly the Mendelian $1:2:1$, but because the heterozygote has its *own* phenotype, the phenotypic ratio is **also** $1:2:1$ (not 3:1). Here R is not completely dominant over r, so Rr is distinguishable from both RR and rr.

Incomplete Dominance — Key Point

The heterozygote shows a **blended/intermediate** phenotype. Mendel's Law of Segregation still holds (alleles do not actually mix), only the Law of Dominance is violated. Phenotypic ratio in $F_2 = 1 : 2 : 1$, same as the genotypic ratio.

3.2 Co-dominance and multiple alleles — the ABO blood group

In **co-dominance** the heterozygote resembles *both* parents simultaneously — both alleles express fully and independently. The classic example is the human ABO blood group, controlled by the gene I with three alleles: I^A, I^B and i.

- I^A produces sugar A on the red-cell surface; I^B produces sugar B; i produces **no** sugar.
- I^A and I^B are each **completely dominant** over i.
- When I^A and I^B are together, *both* sugars appear — this is **co-dominance**, giving blood group AB.

Genotype	Blood group (phenotype)	Dominance type
I^AI^A or I^Ai	A	Complete dominance over i
I^BI^B or I^Bi	B	Complete dominance over i
I^AI^B	AB	Co-dominance
ii	O	Recessive (no sugar)

Because one character is governed by **more than two** alleles (I^A, I^B, i), the ABO system is also the standard example of **multiple alleles**. An individual carries only any two of the three; multiple allelism is observed only at the population level. Three alleles give 6 genotypes and 4 phenotypes (A, B, AB, O).

Real-World Application

ABO blood typing is the everyday application of co-dominance and multiple alleles. Before a transfusion, donor and recipient blood are matched precisely because anti-A and anti-B antibodies in plasma would otherwise clump (agglutinate) mismatched red cells. Group O is the universal red-cell donor; group AB is the universal recipient.

Co-dominance vs Incomplete Dominance

In **incomplete dominance** the heterozygote shows a *new, blended* phenotype (pink). In **co-dominance** the heterozygote shows *both* parental phenotypes *together*, undiluted (group AB has both A and B sugars — not a “mixed” sugar). Do not confuse a blend with a simultaneous joint expression.

4 Inheritance of Two Genes — The Dihybrid Cross

Mendel also crossed plants differing in two characters at once: round–yellow seeds ($RRYY$) with wrinkled–green seeds ($rryy$). The F_1 was uniformly **round and yellow** ($RrYy$), confirming yellow dominant over green and round dominant over wrinkled.

On selfing the F_1, the F_2 showed four phenotypic classes — round yellow, round green, wrinkled yellow, wrinkled green — in the ratio **9:3:3:1**. Crucially, each character on its own still segregated $3:1$ (3 yellow : 1 green; 3 round : 1 wrinkled).

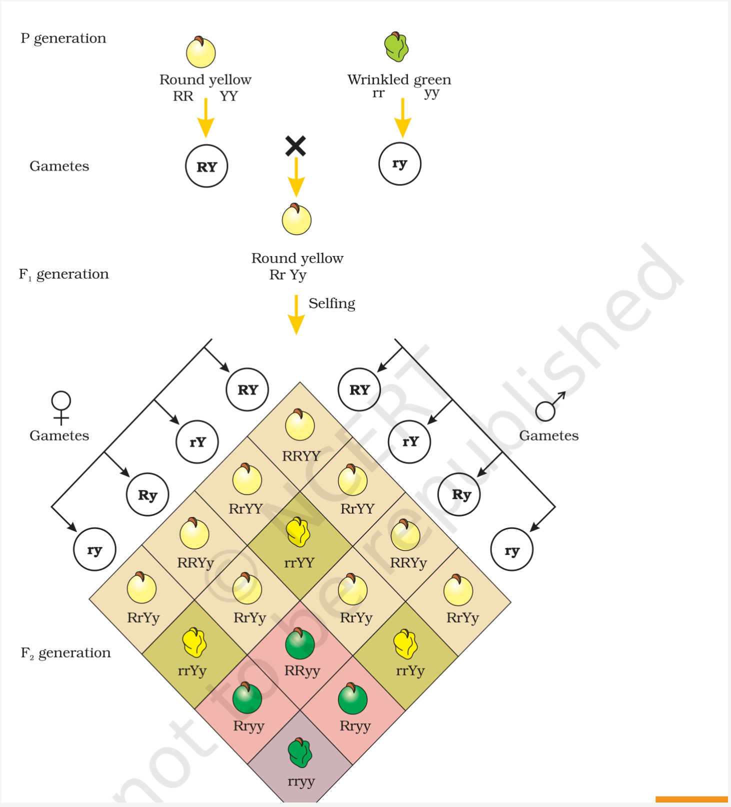


Fig 4.7 — Results of a dihybrid cross between parents differing in two trait-pairs (seed colour and seed shape). The 16-square Punnett grid yields a 9:3:3:1 F_2 phenotypic ratio.

4.1 Law of Independent Assortment

The $RrYy$ F_1 produces **four** gamete types — RY, Ry, rY, ry — each at frequency $\frac{1}{4}$, because the segregation of R/r is independent of the segregation of Y/y. A 4×4 Punnett square (16 boxes) then yields the 9:3:3:1 split.

Dihybrid Ratio as a Product

The 9:3:3:1 ratio is the product of two independent 3:1 monohybrid ratios:

$$(3 \text{ Round} : 1 \text{ Wrinkled}) \times (3 \text{ Yellow} : 1 \text{ Green})$$

$$= 9 \text{ R,Y} : 3 \text{ R,G} : 3 \text{ W,Y} : 1 \text{ W,G}$$

F_2 genotypic ratio $= 1:2:1:2:4:2:1:2:1$ (9 genotypes).

Law of Independent Assortment: *when two pairs of traits are combined in a hybrid, the segregation of one pair of characters is independent of the other pair.* This law holds only for genes on **different chromosomes** (or very far apart on the same one).

Quick Tip

For independently assorting genes, the number of gamete types $= 2^n$ where

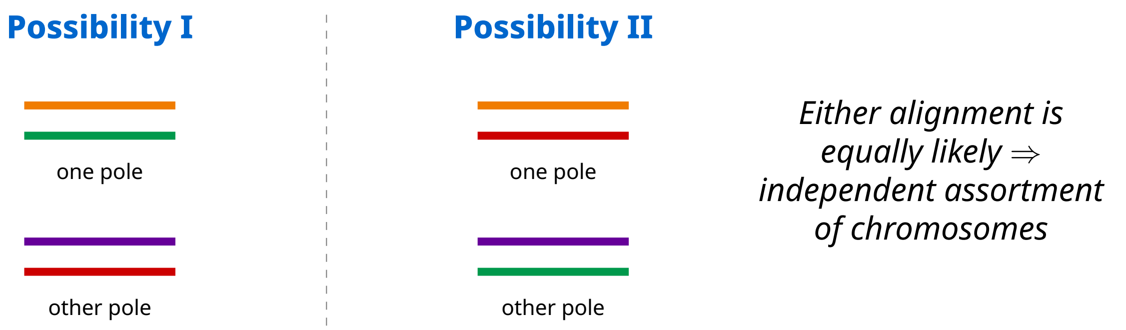
n is the number of heterozygous loci. A $TtRrYy$ plant ($n = 3$) makes $2^3 = 8$ gamete types. This shortcut answers "how many gamete types" questions instantly.

4.2 Chromosomal Theory of Inheritance

Mendel published in 1865 but his work was ignored until 1900, when de Vries, Correns and von Tschermak independently rediscovered it. By then, improved microscopy had revealed **chromosomes** — structures in the nucleus that doubled and divided before each cell division. In 1902, **Walter Sutton** and **Theodor Boveri** noticed that chromosome behaviour during meiosis exactly paralleled Mendel's factors, and united the two ideas into the **Chromosomal Theory of Inheritance**.

Behaviour of Chromosomes	Behaviour of Genes
Occur in pairs	Occur in pairs
Segregate at gamete formation; only one of each pair goes to a gamete	Segregate at gamete formation; only one of each pair goes to a gamete
One pair segregates independently of another pair	Independent pairs segregate independently of each other

The independent alignment of chromosome pairs at the metaphase plate during meiosis I is the physical basis of independent assortment.



The theory was experimentally verified by **Thomas Hunt Morgan** working on the fruit fly *Drosophila melanogaster*. Drosophila was ideal: grown on cheap synthetic medium, a two-week life cycle, large progeny from a single mating, easily distinguishable sexes, and many visible hereditary variations.

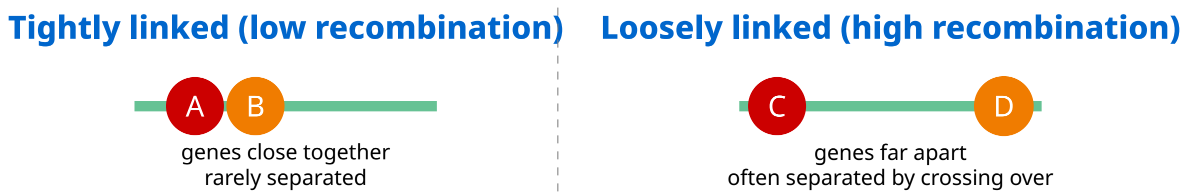
Sutton and Boveri's Insight

The pairing and separation of a pair of homologous chromosomes during meiosis leads to the segregation of the pair of factors (alleles) they carry. Genes are physically located on chromosomes; the two alleles of a gene sit at homologous loci on homologous chromosomes.

4.3 Linkage and Recombination

Morgan crossed yellow-bodied, white-eyed female flies with brown-bodied, red-eyed males and intercrossed the F_1. The F_2 ratio deviated strongly from the expected 9:3:3:1. He found that genes located on the **same chromosome** tend to be inherited *together* — the parental combinations were far more frequent than non-parental ones.

- **Linkage** — the physical association of genes on the same chromosome, so they tend to be inherited together.
- **Recombination** — the generation of non-parental gene combinations (by crossing over during meiosis).
- Genes close together are **tightly linked** (low recombination); genes far apart are **loosely linked** (high recombination). E.g. *white* and *yellow* showed only 1.3% recombination, whereas *white* and *miniature wing* showed 37.2%.



Morgan's student **Alfred Sturtevant** used recombination frequency as a measure of the distance between genes and constructed the first **genetic map**. Genetic maps are still the starting point for whole-genome sequencing, as in the Human Genome Project.

> **Real-World Application**
>
> Recombination-frequency mapping is the conceptual ancestor of modern gene mapping used to locate disease genes. Linkage analysis in human families helped pinpoint the chromosomal locations of genes for cystic fibrosis, Huntington's disease and many cancers, guiding genetic counselling and diagnostic testing.

5 Polygenic Inheritance and Pleiotropy

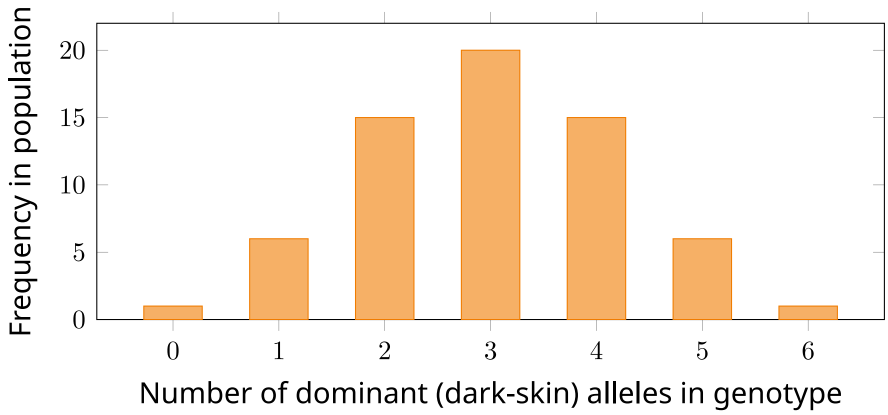
5.1 Polygenic inheritance

Mendel studied traits with sharp alternatives. But many traits — human height, skin colour — vary continuously across a gradient rather than in distinct classes. Such traits are **polygenic**: controlled by three or more genes, with each allele contributing additively, and additionally influenced by the **environment**.

Take human skin colour as the model, controlled (in this simplified scheme) by three genes A, B, C. Dominant alleles (A, B, C) add darkness; recessive alleles (a, b, c) give lighter skin.

- $AABBCC$ (6 dominant alleles) — darkest skin.
- $aabbcc$ (0 dominant alleles) — lightest skin.
- Intermediate numbers of dominant alleles give a graded spectrum of intermediate shades.

The phenotype reflects the **total number of contributing (dominant) alleles**, not which specific genes carry them. This additive effect, plus environmental input (e.g. sun exposure), produces the smooth bell-shaped distribution typical of quantitative traits.



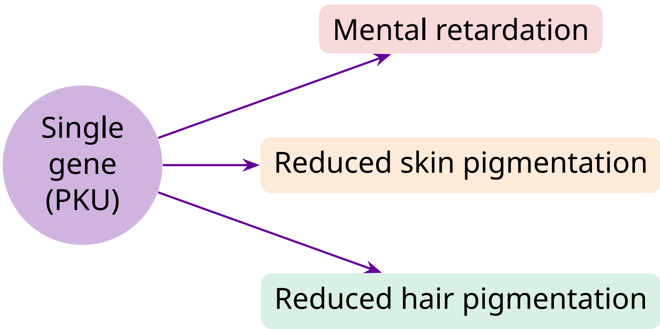
Polygenic Inheritance — Key Features

(1) Multiple genes (≥ 3) act on one character. (2) Each allele has a small **additive** effect. (3) The environment modifies the phenotype. (4) The result is **continuous (quantitative) variation** with a normal-distribution spread, not discrete classes.

5.2 Pleiotropy

The reverse situation also occurs: a single gene that influences **multiple** phenotypic traits is called a **pleiotropic gene**. The usual mechanism is that the gene acts on a metabolic pathway that feeds into several phenotypes.

Phenylketonuria (PKU) is the textbook example. A mutation in the gene for the enzyme *phenylalanine hydroxylase* produces several effects at once: mental retardation *and* reduced hair and skin pigmentation. One gene mutation, many phenotypic consequences.



Poly vs Pleio

Polygenic = *poly* (many) genes → *one* trait (continuous, e.g. skin colour). **Pleio**tropic = *one* gene → *many* traits (e.g. PKU, sickle-cell anaemia). Read the arrow direction: many→one is polygenic; one→many is pleiotropic.

6 Sex Determination

The chromosomal basis of sex was first hinted at by **Henking (1891)**, who traced a nuclear structure he called the "X body" through insect spermatogenesis; only 50% of sperm received it. This was later recognised as the **X-chromosome**. Chromosomes that differ between the sexes are **sex chromosomes**; the rest are **autosomes**.

6.1 XO, XY and ZW systems

System	Mechanism	Example
XO type	Female XX; male XO (one X, no second sex chromosome). Male heterogametic.	Grasshopper, many insects
XY type	Female XX; male XY. Both sexes have same chromosome number. Male heterogametic.	Humans, *Drosophila*
ZW type	Female ZW; male ZZ. **Female** heterogametic.	Birds

In **male heterogamety** (XO, XY) the male makes two kinds of gamete; in **female heterogamety** (ZW) the female makes two kinds of gamete. The heterogametic sex determines the offspring's sex.

Humans (XY type)

Female XX × Male XY

egg: X   sperm: X or Y

↓

X + X → **XX (girl)**   X + Y → **XY (boy)**

Sperm decides the sex — 50:50 each pregnancy

Birds (ZW type)

Female ZW × Male ZZ

egg: Z or W   sperm: Z

↓

Z+Z → ZZ (male)
W+Z → ZW (female)

Egg decides the sex

6.2 Sex determination in humans

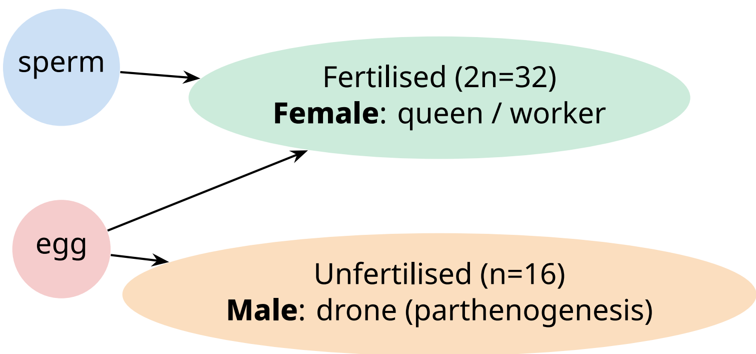
Humans have 23 pairs of chromosomes: 22 pairs of autosomes plus one pair of sex chromosomes. Females are XX; males are XY. During spermatogenesis, 50% of sperm carry an X and 50% carry a Y; every ovum carries an X. Fertilisation by an X-bearing sperm gives a girl (XX); a Y-bearing sperm gives a boy (XY).

The Sperm Determines the Child's Sex

Because the mother contributes only X, it is the **father's sperm** (X or Y) that decides the sex of the child, with an equal 50% probability of either sex in each pregnancy. Blaming women for the sex of a child is biologically baseless.

6.3 Sex determination in honey bee — haplodiploidy

Honey bees use a **haplodiploid** system based on the number of chromosome *sets*, not specific sex chromosomes. A fertilised egg (diploid, 32 chromosomes) develops into a **female** (queen or worker). An unfertilised egg (haploid, 16 chromosomes) develops into a **male (drone)** by **parthenogenesis**.



Quick Tip

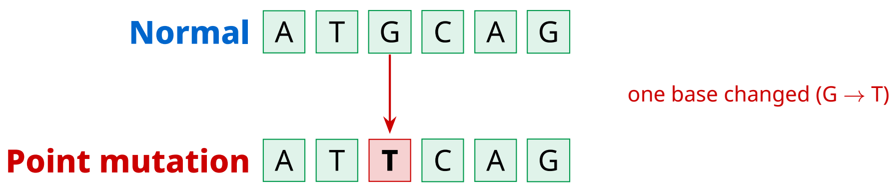
Remember the drone's odd family tree: a male honey bee has **no father** (from an unfertilised egg) and **cannot have sons**, but he **has a grandfather and can have grandsons**. Drones make sperm by **mitosis** (they are already haploid). This is a recurring NEET one-liner.

7 Mutation

Mutation is a change in the DNA sequence, leading to altered genotype and phenotype. Alongside recombination, it is a major source of variation.

- **Chromosomal mutations / aberrations** — loss (*deletion*) or gain (*insertion / duplication*) of a DNA segment alters the chromosome. Common in cancer cells.
- **Point mutation** — change in a single base pair. The classic example is **sickle-cell anaemia** (a single GAG→GUG change).
- **Frame-shift mutation** — deletion or insertion of base pairs that shifts the reading frame (detailed in Chapter 5).

Mutagens are physical/chemical agents that induce mutations — for example, **UV radiation** is a mutagen.



Mutation in One Line

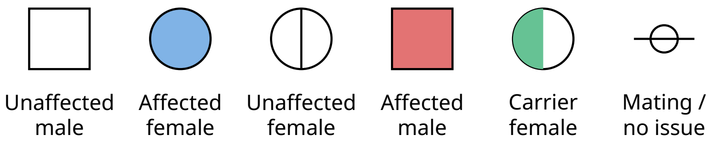
Mutation = any heritable change in the genetic material. A **point mutation** alters a single base pair; **chromosomal mutations** alter chromosome structure or number. Mutations are the raw material of variation and, ultimately, evolution.

8 Genetic Disorders

A number of human disorders are caused by inherited altered genes or chromosomes. They fall into two broad classes: **Mendelian disorders** (single-gene mutations) and **Chromosomal disorders** (changes in chromosome number/structure).

8.1 Pedigree analysis

Because controlled crosses are impossible in humans, geneticists study the **family history** of a trait across several generations. This **pedigree analysis** represents the trait's inheritance as a family tree and reveals whether a trait is dominant or recessive, and whether it is autosomal or sex-linked.



Standard symbols (after Fig 4.13): squares = males, circles = females, filled = affected, half-shaded = carrier, horizontal line = mating.

8.2 Mendelian disorders

These arise from a mutation in a **single gene** and are transmitted just like Mendel's traits; pedigree analysis traces the pattern. Common examples: haemophilia, cystic fibrosis, sickle-cell anaemia, colour blindness, phenylketonuria, thalassemia. They may be dominant or recessive, and autosomal or sex-linked.

Colour blindness — an **X-linked recessive** defect in red or green cones, causing failure to distinguish red and green. It affects ≈ 8% of males but only ≈ 0.4% of females, because males have a single X. A carrier mother's son has a 50% chance of being colour blind; she herself is unaffected as her second X carries the dominant normal allele.

Haemophilia — an **X-linked recessive** disease in which a clotting-cascade protein is defective, so even a minor cut causes prolonged bleeding. A carrier (heterozygous) female transmits it to some sons. An affected female is extremely rare (her father would have to be haemophilic and her mother at least a carrier). The pedigree of Queen Victoria famously carried haemophilia through European royalty.

Sickle-cell anaemia — an **autosomal recessive** disorder; the allele pair is Hb^A and Hb^S. Only $Hb^S Hb^S$ homozygotes are diseased; $Hb^A Hb^S$ heterozygotes are unaffected **carriers** (sickle-cell trait). The defect is a single base substitution at the 6^{th} codon of the β-globin gene (GAG→GUG), replacing **glutamic acid by valine** at the 6^{th} position of the β-chain. Under low oxygen, mutant haemoglobin polymerises and the biconcave RBC distorts into a sickle shape. This is also a clear case of **pleiotropy** and of **point mutation**.

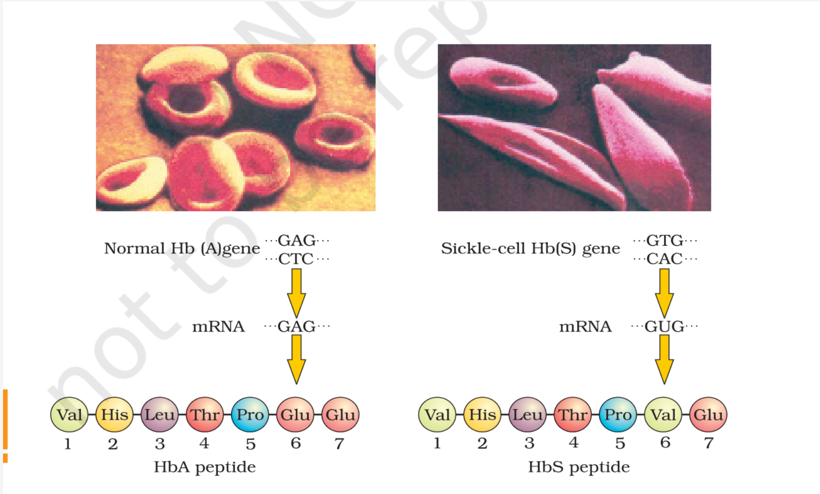


Fig 4.15 — RBC micrographs and the β-globin amino-acid sequence: (a) normal individual (Glu at position 6); (b) sickle-cell individual (Val at position 6, from a single GAG→GUG base change).

Phenylketonuria — an **autosomal recessive** inborn error of metabolism. The affected individual lacks the enzyme that converts phenylalanine to tyrosine, so phenylalanine accumulates, is converted to phenylpyruvic acid and derivatives that accumulate in the brain (mental retardation) and are excreted in urine.

Thalassemia — an **autosomal recessive** blood disorder. Mutation/deletion reduces the synthesis of a globin chain. In α**-thalassemia** the α-chain is affected (genes *HBA1*, *HBA2* on chromosome 16); in β**-thalassemia** the β-chain is affected (gene *HBB* on chromosome 11).

Thalassemia vs Sickle-cell Anaemia

Both are autosomal recessive blood disorders, but they differ in nature. Thalassemia is a **quantitative** problem — too few globin molecules are made. Sickle-cell anaemia is a **qualitative** problem — a normal *amount* of an *incorrectly functioning* globin is made. Examiners test this exact distinction.

8.3 Chromosomal disorders

These are caused by the **absence, excess, or abnormal arrangement** of one or more chromosomes.

- **Aneuploidy** — failure of chromatid segregation gives gain or loss of a chromosome. *Trisomy* = one extra chromosome; *monosomy* = one missing chromosome.
- **Polyploidy** — failure of cytokinesis adds an entire extra chromosome set; common in plants.

Down's syndrome is caused by trisomy of chromosome 21, so the total chromosome number becomes 47. It was first described by Langdon Down (1866). Features: short stature, small round head, furrowed/wrinkled tongue, partially open mouth, broad palm with a characteristic palm crease, congenital heart disease, and retarded physical, psychomotor and mental development.

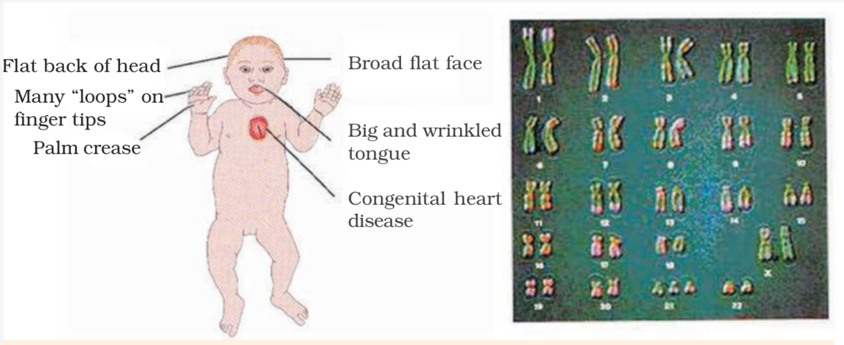


Fig 4.16 — An individual with Down's syndrome (trisomy 21) and the corresponding karyotype showing three copies of chromosome 21.

Klinefelter's syndrome — an extra X giving karyotype $47,\ XXY$. Overall masculine build but with feminine development (gynaecomastia — breast development); such individuals are sterile.

Turner's syndrome — absence of one X, karyotype $45,\ X0$. Such females are sterile, ovaries are rudimentary, and other secondary sexual characters are lacking.

Disorder	Karyotype	Key features
Down's syndrome	47 (trisomy 21)	Short stature, furrowed tongue, palm crease, congenital heart defect, mental retardation
Klinefelter's syndrome	47, XXY	Masculine build with gynaecomastia; sterile
Turner's syndrome	45, X0	Sterile female; rudimentary ovaries; lacking secondary sexual characters

Real-World Application

Chromosomal disorders are detected before birth by **karyotyping** of fetal cells (from amniocentesis or chorionic villus sampling). Counting and arranging the chromosomes reveals trisomies (Down's) or sex-chromosome anomalies (Klinefelter's, Turner's), informing prenatal genetic counselling.

Related Collegedunia Resources

Same chapter — other resources:
- NCERT Solutions
- Formula Sheet
- NCERT Book PDF
- Exemplar Book PDF
- Exemplar Solutions
- Handwritten Notes

Continue learning:
- Ch 3: Reproductive Health
- Ch 5: Molecular Basis of Inheritance
- Class 12 Biology — All Chapters

9 Quick Reference Summary

9.1 Mendel's laws at a glance

Law	Statement
Law of Dominance	In a dissimilar factor pair, one (dominant) masks the other (recessive); explains the 3:1 F_2 ratio.
Law of Segregation	Alleles segregate during gamete formation; each gamete gets only one. No exceptions (Law of Purity of Gametes).
Law of Independent Assortment	Segregation of one trait-pair is independent of another (for genes on different chromosomes); gives 9:3:3:1.

9.2 Standard genetic ratios

Ratios to Memorise

- Monohybrid F_2: phenotype $3:1$ genotype $1:2:1$
- Incomplete dominance F_2: phenotype $=$ genotype $= 1:2:1$
- Dihybrid F_2: phenotype $9:3:3:1$ genotype $1:2:1:2:4:2:1:2:1$
- Test cross of heterozygote: $1:1$ (dominant : recessive)
- Gamete types from n heterozygous loci $= 2^n$

9.3 Inheritance patterns of key disorders

Disorder	Inheritance	Defect
Colour blindness	X-linked recessive	Defective red/green cones
Haemophilia	X-linked recessive	Defective blood-clotting protein
Sickle-cell anaemia	Autosomal recessive	GAG→GUG; Glu→Val at β-6 (point mutation)
Phenylketonuria	Autosomal recessive	Lacks phenylalanine hydroxylase
Thalassemia	Autosomal recessive	Reduced α- or β-globin synthesis
Down's syndrome	Trisomy 21 (chromosomal)	Extra chromosome 21 (47 total)
Klinefelter's syndrome	47, XXY (chromosomal)	Extra X chromosome
Turner's syndrome	45, X0 (chromosomal)	Missing X chromosome

9.4 Sex-determination systems

System	Heterogametic sex	Example
XO	Male (XX female / XO male)	Grasshopper
XY	Male (XX female / XY male)	Humans, *Drosophila*
ZW	Female (ZW female / ZZ male)	Birds
Haplodiploidy	— (2n female / n male)	Honey bee

Exam Focus for Boards and NEET

The highest-yield items from this chapter: working out monohybrid/dihybrid Punnett squares; distinguishing incomplete dominance vs co-dominance; ABO blood-group genetics (genotypes from phenotypes); pedigree interpretation (dominant vs recessive, autosomal vs sex-linked); and the karyotypes of Down's, Klinefelter's and Turner's syndromes. Practise blood-group problems and pedigree problems until they are automatic.

End of Chapter 4 — Principles of Inheritance and Variation. Revise the Quick Reference tables before the exam.