

Inheritance and variation

3.1 Chromosomes and Mechanism of Inheritance:

Inheritance

Passing genetic information from one generation to the next is called **heredity** or **inheritance**. The process of inheritance was studied and understood even before chromosomes were seen or genes were discovered.

Gregor Mendel (Moravia, 1822)

Mendel was the first to correctly explain the mechanism of inheritance using the hybridisation technique. Using statistical methods, Mendel studied 7 traits in the Garden Pea plant (*Pisum sativum*). Mendel proposed the principles of heredity, which later became the fundamental laws of inheritance as confirmed by Correns in 1900.

He suggested that traits are not passed on physically, but through certain elements present in the gametes. Mendel called these elements “**factors**”, which control the expression of specific traits. He explained that these factors are discrete units, now known as **genes**. They exist in pairs in parents and separate from each other during gamete formation without mixing.

Reasons for Mendel's Success:

1. He designed his **experiments very carefully** and used **large sample sizes** for accuracy.
2. He recorded detailed observations, counted the number of plants of each type, and **analysed results using ratios**.
3. In pea plants, **contrasting characters** are **easy to identify**.
4. Each of the **seven traits** studied in pea plants was controlled by a **single factor**, located on **different chromosomes**, and passed on from one generation to the next.
5. He **introduced** the important **concepts of dominance and recessiveness** in inheritance.

3.2 Genetic Terminology:

1. **Character:** A **specific feature** of an organism, e.g., height of the stem in a plant.
2. **Trait:** A **detectable form or variant of a character** that is inherited, e.g., tall or dwarf.
3. **Factor:** A **unit of heredity** present in an organism that **controls the inheritance and expression of a character**. These factors are **passed from one generation to the next through gametes**. Today, these factors are known as **genes**.
4. **Gene:** A **specific segment of DNA** that **controls the inheritance and expression of a character**.
5. **Alleles (Allelomorphs):** **Different alternative forms of the same gene** are called alleles. They are **found at the same position (locus) on homologous chromosomes**. Controls the trait of the character; the term "*allele*" is a shortened form of "*allelomorph*."
6. **Dominant:** An **allele that expresses itself** even when **paired with a different allele (heterozygous condition)**. It is the allele that **expresses itself in the F₁ generation and masks the effect** of the other allele.
7. **Recessive:** An allele that **remains unexpressed in the presence of a dominant allele (heterozygous condition)**. It can only **express** when **two identical copies** are present (**homozygous condition**). This allele does **not appear** in the **F₁ hybrid**.
8. **Phenotype:** The **observable (external) physical appearance** of an individual for a **particular trait**. The **combination of alleles determines it**.
Example: In pea plants, **tall and dwarf are two phenotypes**. Tall may result from **TT** or **Tt**, while dwarf results from **tt**.
9. **Genotype:** The **genetic constitution or makeup of an organism** with respect to a **trait**. It represents the **allele combination** an individual carries.
Example: A tall pea plant may have genotype **TT** or **Tt**, while a dwarf plant has **tt**.
10. **Homozygous (pure):** An individual having **identical alleles** for a trait is called homozygous. It **breeds true** for that trait and **produces only one type of gamete**.
Example: Tall (**TT**) or dwarf (**tt**).

11. **Heterozygous:** An individual having **contrasting alleles** for a trait is called heterozygous. It does **not breed true** and produces **two types of gametes**. Such individuals are also called **hybrids**.
Example: F_1 generation hybrid **Tt** in pea plants.
12. **Pure Line:** A group of individuals that are **homozygous** and **true-breeding** for **one or more traits**. They **consistently pass** the **same character** to the next generation. Such a line **originates** from a **single homozygous parent** through **self-fertilisation**.
13. **Monohybrid:** An individual **heterozygous for a single trait**, produced by **crossing two pure parents** that **differ** in one pair of **contrasting characters**.
Example: A hybrid tall plant (**Tt**) obtained from a cross between pure tall (**TT**) and pure dwarf (**tt**) parents.
14. **F₁ Generation:** Latin word *filius* (son) or *filia* (daughter). The **first filial generation** consists of all **offspring produced from a cross between two pure parents** with **contrasting traits**. It represents the first generation from such a mating.
15. **F₂ Generation:** The **second filial generation** produced by **self-fertilisation (inbreeding)** of F_1 offspring.
Example: Crossing two **F₁ hybrids (Tt × Tt)** produces the F_2 generation.
16. **Punnett Square (Checkerboard):** A **probability table** that shows all possible combinations of gametes from opposite parents. It is a diagram used to predict the types and ratios of offspring from a genetic cross.
17. **Homologous Chromosomes:** Chromosomes that are similar in size, structure, and genetic content, found in diploid cells. They pair (synapse) during meiosis.
18. **Back Cross:** A cross between an F_1 hybrid and either of its parents.
Example: **Tt × TT** (pure tall) or **Tt × tt** (pure dwarf).
19. **Test Cross:** A cross between an F_1 hybrid and a homozygous recessive parent, used to determine whether the hybrid is homozygous or heterozygous.
Example: **Tt × tt**.
20. **Phenotypic ratio:** It is the ratio of the offspring produced in F_2 and subsequent generations with respect to their physical appearance, e.g. 3Tall: 1 dwarf, is F_2 'Phenotypic ratio' in a monohybrid cross.

21. **Genotypic ratio:** It is the ratio of the offspring produced in the F₂ and subsequent generations with respect to their genetic makeup, e.g. 1 TT : 2Tt : 1 tt, is F₂ genotypic ratio in a monohybrid cross.
22. **Monohybrid cross:** A cross between parents which **differ in only one heritable trait** is called a monohybrid cross. e.g. **cross of pure tall and pure dwarf** plants. Mendel performed the monohybrid cross between two pea plants with **only one pair of contrasting** characters.
23. **Dihybrid cross:** A cross between parents **differing in two heritable traits** is called a dihybrid cross, e.g. cross of a pure tall, round-seeded plant with a dwarf, wrinkled-seeded plant. Mendel also performed the dihybrid cross between pea plants that differed in two pairs of contrasting characters.

Punnett Square: TtRr × TtRr

	TR	Tr	tR	tr
TR	TTRR	TTRr	TtRR	TtRr
Tr	TTRr	TTrr	TtRr	Tttr
tR	TtRR	TtRr	ttRR	ttRr
tr	TtRr	Tttr	ttRr	tttr

Genotypes — samples (from the 16 boxes)

TTRR : 1: TTRr : 2: TTrr : 1 : TtRR : 2 : TtRr : 4: Tttr : 2 : ttRR : 1: ttRr : 2 : tttr : 1

Genotypic ratio

1 : 2 : 1 : 2 : 4 : 2 : 1 : 2 : 1

Phenotypes — counts & ratio

Dominance rules: T₋ = Tall, tt = dwarf; R₋ = Round, rr = wrinkled

Tall, Round (T₋ R₋) = 9/16

(TTRR + TTRr + TtRR + TtRr → 1 + 2 + 2 + 4 = 9)

Tall, Wrinkled (T₋ rr) = 3/16

(TTrr + Tttr → 1 + 2 = 3)

Dwarf, Round (tt R₋) = 3/16

(ttRR + ttRr → 1 + 2 = 3)

Dwarf, Wrinkled (tt rr) = 1/16

(tttr → 1)

Phenotypic ratio

9 : 3 : 3 : 1

(Tall-Round : Tall-Wrinkled : Dwarf-Round : Dwarf-Wrinkled)

Table 1 Summary of Basic Genetic Terminology with Definitions and Examples:

Terminology	Definition	Example
Character	A specific feature of an organism.	Height of stem in <i>Pisum sativum</i> .
Trait	Detectable form/variant of a character.	Tall or dwarf stem.
Factor	A unit of heredity controlling a character (now called a gene).	Mendel's "factors" for height.
Gene	Segment of DNA controlling inheritance & expression of a character.	Gene for stem height.
Alleles	Alternative forms of the same gene, present at the same locus on homologous chromosomes.	T (tall) and t (dwarf).
Dominant	Allele that expresses even in heterozygous condition.	T (tall) masks t (dwarf).
Recessive	Allele that expresses only when identical alleles are present.	tt = dwarf pea plant.
Phenotype	Physical expression of a trait determined by combination of genotype.	Tall or dwarf pea plants.
Genotype	Genetic makeup of an organism for a trait.	TT, Tt, or tt.
Homozygous	Condition with identical alleles for a trait.	TT (tall) or tt (dwarf).
Heterozygous	Condition with contrasting alleles for a trait.	Tt (tall hybrid).
Pure Line	Population of true-breeding individuals passing same character.	Self-fertilized TT or tt plants.
Monohybrid	Individual heterozygous for one trait.	Tt (from TT × tt cross).
F₁ Generation	First generation from cross of two pure parents.	TT × tt → all Tt (tall).

F₂ Generation	Second generation from selfing F ₁ hybrids.	$Tt \times Tt \rightarrow 1 TT : 2 Tt : 1 tt$.
Punnett Square	A diagrammatic representation showing all gamete combinations in a cross.	$Tt \times Tt$ checkerboard.
Homologous Chromosomes	Chromosomes similar in size, structure, genes; synapse in meiosis.	Human chromosome pair 1.
Back Cross	Cross between an F ₁ hybrid and one of its parents.	$Tt \times TT$ or $Tt \times tt$.
Test Cross	Cross between an F ₁ hybrid and a homozygous recessive to test the genotype.	$Tt \times tt$.
Phenotypic Ratio	Ratio of offspring by physical traits.	3 tall: 1 dwarf.
Genotypic Ratio	Ratio of offspring by genetic makeup.	1 TT : 2 Tt : 1 tt.
Monohybrid Cross	Cross between parents differing in one trait.	Tall (TT) $\times$ Dwarf (tt).
Dihybrid Cross	Cross between parents differing in two traits.	Tall round (TTRR) $\times$ dwarf wrinkled (ttrr).

Table 2: Summary of Basic Genetic Terminology with Key words, Definitions and Examples:

Terminology	Category (Nature)	Key words	Example
Character	Feature	Feature, Organism	Height of stem in pea plant.
Trait	Feature	Variant, Inherited	Tall or dwarf stem.
Factor	Unit	Heredity, Control	Mendel's "factors" for height.
Gene	Unit	DNA, Expression	Gene for stem height.
Alleles	Unit	Alternative, Locus	T (tall) and t (dwarf).
Dominant	Property	Express, Mask	T (tall) masks t (dwarf).

Recessive	Property	Hidden, Homozygous	tt = dwarf pea plant.
Phenotype	Expression	Appearance, Observable	Tall or dwarf pea plants.
Genotype	Expression	Genetic, Constitution	TT, Tt, or tt.
Homozygous	Condition	Identical, True-breeding	TT (tall) or tt (dwarf).
Heterozygous	Condition	Contrasting, Hybrid	Tt (tall hybrid).
Pure Line	Population	Homozygous, Consistent	Self-fertilized TT or tt plants.
Monohybrid	Condition	Single, Cross	Tt (from TT × tt cross).
F₁ Generation	Population	Filial, Offspring	TT × tt → all Tt (tall).
F₂ Generation	Population	Selfing, Second	Tt × Tt → 1 TT : 2 Tt : 1 tt.
Punnett Square checkerboard	diagrammatic representation	Probability, Diagram	Tt × Tt checkerboard.
Homologous Chromosomes	Unit	Pair, Similar	Human chromosome pair 1.
Back Cross	Experiment	Parent, Hybrid	Tt × TT or Tt × tt.
Test Cross	Experiment	Recessive, Identify	Tt × tt.
Phenotypic Ratio	Ratio	Physical, Proportion	3 tall : 1 dwarf.
Genotypic Ratio	Ratio	Genetic, Proportion	1 TT : 2 Tt : 1 tt.
Monohybrid Cross	Experiment	One, Contrasting	Tall (TT) × Dwarf (tt).
Dihybrid Cross	Experiment	Two, Contrasting	Tall round (TTRR) × dwarf wrinkled (ttrr).

Table 3. Mendel's Seven Characters in *Pisum sativum* with Their Contrasting Traits and Genotypes:

Character	Dominant Trait	Genotype (Dominant)	Recessive Trait	Genotype (Recessive)
Seed shape	Round	RR	Wrinkled	rr
Seed colour	Yellow	YY	Green	yy
Flower colour	Violet/Purple	PP	White	pp
Pod shape	Inflated	II	Constricted	ii
Pod colour	Green	GG	Yellow	gg
Flower position	Axial	AA	Terminal	aa
Stem length	Tall	TT	Dwarf	tt

3.3 Mendel's Laws of Inheritance :

1. Law of Dominance

Statement:

“When two homozygous individuals with one or more sets of contrasting characters are crossed, the alleles (characters) that appear in F₁ are dominant and those which do not appear in F₁ are recessive”.

Explanation:

- In a pair of contrasting factors (alleles), one is **dominant** and the other is **recessive**.
- The dominant allele expresses itself in the **F₁ generation**, while the recessive allele is masked.

- In **F₂ generation**, both alleles reappear in a specific ratio (3:1 in a monohybrid cross).
- **Example:**

Parent cross: $TT \times tt$

Gametes: $T \times t$

F₁ : Tt

F₁ Result: All **Tt (Tall)** → Dominant trait expressed.

2. Law of Segregation (Law of Purity of Gametes)

Statement:

“When hybrid (F₁) forms gametes, the alleles segregate from each other and enter in different gametes”. The gametes formed are pure in that they carry only one allele each (either dominant allele or recessive allele). Hence, this law is also described as “Law of purity of gametes”.

Explanation:

- Alleles **do not mix** or blend.
- During gamete formation, the two alleles of a trait **separate** and each gamete gets only **one allele** (pure).
- Explains why the recessive trait reappears in **F₂ generation**.

Example:

F₁ Parent: $Tt \times Tt$

Gametes: $T, t \times T, t$

F₂ :

	T	t
T	TT	Tt
t	Tt	tt

F₂ Result:

- Genotype ratio = **1 TT : 2 Tt : 1 tt**
- Phenotype ratio = **3 Tall : 1 Dwarf**

3. Law of Independent Assortment :

(Assort: To arrange or to divide into categories)

Statement:

“When hybrid possessing two (or more) pairs of contrasting factors (alleles) forms gametes, the factors in each pair segregate independently of the other pair”.

- This is the basic principle of genetics based on **a dihybrid** cross given by Mendel.
- Alleles of different genes (located on different chromosomes) **assort independently** during gamete formation.
- Results in new combinations of traits.
- **F₂ phenotypic ratio: 9:3:3:1**

(Dihybrid Cross: TtYy × TtYy)

Parent cross: **TtYy × TtYy**

Gametes: **TY, Ty, tY, ty × TY, Ty, tY, ty**

	TY	Ty	tY	ty
TY	TTYy	TTYy	TtYY	TtYy
Ty	TTYy	TTyy	TtYy	Ttyy
tY	TtYY	TtYy	ttYY	ttYy
ty	TtYy	Ttyy	ttYy	ttyy

Result :

Phenotypic ratio: 9:3:3:1

Genotypic ratio: 1:2:1:2:4:2:1:2:1

Table 4: Comparison of the Mendel's Laws:

Aspect	Law of Dominance	Law of Segregation	Law of Independent Assortment
Statement	When two homozygous individuals with one or more sets of contrasting characters are crossed, the alleles (characters) that appear in F ₁ are dominant and those which do not appear in F ₁ are recessive".	"When two homozygous individuals with one or more sets of contrasting characters are crossed, the alleles (characters) that appear in F ₁ are dominant and those which do not appear in F ₁ are recessive".	"When hybrid possessing two (or more) pairs of contrasting factors (alleles) forms gametes, the factors in each pair segregate independently of the other pair".
Cross Example	Monohybrid cross (Tt × Tt) shows tall plants dominate over dwarf.	Monohybrid cross (Tt × Tt) → offspring ratio follows 3:1 phenotype, 1:2:1 genotype.	Dihybrid cross (TtYy × TtYy) → offspring ratio follows 9:3:3:1 in F ₂ generation.
Phenotypic Expression	Only the dominant trait is expressed in F ₁ generation.	Both parental traits reappear in F ₂ due to separation of alleles.	New combinations of traits appear in F ₂ (recombinants).
Generation Focus	Explains F ₁ expression.	Explains F ₂ segregation pattern.	Explains F ₂ new combinations and ratios.
Ratio Produced	F ₁ all show dominant trait.	F ₂ → Phenotypic ratio 3:1, Genotypic ratio 1:2:1.	F ₂ → Dihybrid ratio 9:3:3:1.

3.4 Back Cross and Test Cross

a. Back Cross

- F_1 individuals are usually selfed to obtain F_2 progeny.
- They can also be crossed with one of the **original parents** (either recessive or dominant).
- Such a cross is called a **Back Cross**.

b. Test Cross

- Crossing of an **F_1 hybrid** with its **homozygous recessive parent**.
- Purpose: To **determine genotype** of an individual with dominant expression.
- Features:
 - Simple, repeatable, and predictable.
 - Distinguishes **homozygous** (pure) from **heterozygous** (hybrid).

Example (Pea plant – Flower Colour): Case 1: F_1 is Homozygous (PP)

 	P	P
P	Pp	Pp
P	Pp	Pp

All offspring = Violet → Parent is **Homozygous**

Case 2: F_1 is Heterozygous (Pp)

 	P	p
p	Pp	pp
p	Pp	pp

Offspring ratio = **1 Violet : 1 White** → Parent is **Heterozygous**

Comparison: Back Cross vs Test Cross

Feature	Back Cross	Test Cross
Definition	Crossing F_1 with any one of the two parents (dominant or recessive).	Crossing F_1 with homozygous recessive parent .
Purpose	To study inheritance and transfer parental traits.	To determine whether an individual is homozygous or heterozygous.
Types	Two types: with dominant parent / with recessive parent.	Only one type: always with recessive parent.
Result	Depends on the parent used (all dominant traits if crossed with dominant parent; segregation if recessive parent is used).	100% dominant (homozygous) OR 1:1 ratio of dominant : recessive (heterozygous).
Use	Used in crop improvement and breeding experiments.	Used in genetic analysis, identification of genotype.

Neo-Mendelism

3.5 Deviations from Mendel's Findings (Neo-Mendelism)

Mendel's Generalisations:

1. **Single trait** → **single gene** → **two alleles**.
2. One allele is **completely dominant** over the other.
3. Genes for different traits assort **independently** (on different chromosomes).

Later Observations (Deviations):

- Post-Mendelian studies showed **exceptions** to these rules.

- These are described as **Neo-Mendelism**, adding further understanding of inheritance.

Gene Interactions:

The expression of a gene is not always independent; it can be **modified or influenced by another gene**. Such relationships are called **gene interactions**, which are of two main types:

I. Intragenic Interactions

- Occur **between alleles of the same gene**.
- Examples:
 - Incomplete dominance** – neither allele is completely dominant, producing an intermediate phenotype.

Definition:

Incomplete dominance is a type of inheritance in which **neither allele of a gene** is **completely dominant** over the other, and both alleles **express** themselves **partially**. As a result, the **heterozygous condition produces an intermediate phenotype** that is different from both homozygous conditions.

- **Key Points:**
 - The trait in the heterozygote appears as a **blend or mixture** of the two parental traits.
 - This does **not mean the alleles have mixed or lost their identity; they can separate again** in the next generation.
 - Example: *Mirabilis jalapa* (Four O'Clock plant) – Red (RR) × White (rr) → Pink (Rr).

Cross: Red (RR) × White (rr)

F₁ Generation: All Pink (Rr)

F₂ Generation (Rr × Rr):

R	r	
+-----+-----+		
R		RR Rr
+-----+-----+		
r		Rr rr
+-----+-----+		

- **Genotype ratio:** 1 RR : 2 Rr : 1 rr

- **Phenotype ratio:** 1 Red : 2 Pink : 1 White

b. **Co-dominance** – both alleles express equally in the heterozygote.

Definition:

Co-dominance is a type of inheritance in which **both alleles of a gene pair are fully expressed and independently visible** in the heterozygous condition, without blending. Thus, the **F₁ hybrid exhibits both parental traits side by side**.

• **Key Points:**

- Both alleles are **equally dominant**, and neither masks the other.
- The phenotype of the heterozygote shows the effect of **both alleles simultaneously**.
- Example: Human **AB blood group** – genotype $I^A I^B$ shows the presence of **both A and B antigens** on red blood cells.

Cross: Blood group A ($I^A I^A$) × Blood group B ($I^B I^B$)

F₁ Generation: All AB ($I^A I^B$)

Punnett Square ($I^A \times I^B$):

	I^A	I^A
	+-----+	+-----+
I^B	$I^A I^B$	$I^A I^B$
	+-----+	+-----+
I^B	$I^A I^B$	$I^A I^B$
	+-----+	+-----+

- All offspring = **AB blood group** (both antigens expressed equally).

Feature	Incomplete Dominance	Co-dominance
Definition	Both alleles express partially; neither is completely dominant.	Both alleles express equally and independently.
F₁ Hybrid Expression	Shows an intermediate phenotype (blending).	Shows both parental traits together (no blending).
Phenotypic Ratio in F₂	1 : 2 : 1 (same as genotypic ratio).	No fixed ratio (depends on alleles involved).

Example	<i>Mirabilis jalapa</i> flower color: Red (RR) × White (rr) → Pink (Rr).	Human AB blood group : $I^A I^B$ genotype → both A and B antigens expressed.
Key Point	Expression is mixed (intermediate).	Expression is side-by-side (both traits visible).

- **Multiple alleles** – more than two alternative forms of a gene, e.g., human blood groups.

ii. Intergenic (Non-allelic) Interactions

- Occur **between alleles of different genes** located on the **same or different chromosomes**.

1. Multiple Alleles

- **Definition:** When a single gene has more than **two alternative forms (alleles)**, they are called *multiple alleles*.
- Although more than two alleles exist in the population, an **individual can possess only two** (diploid condition).
- Discovered first in the coat colour of rabbits.

Table 3.3 : Few phenotypes and genotypes in *Drosophila*

Phenotype	Genotype
Normal wings 	vg^+
Nicked wings 	vg^{ni}
Notched wings 	vg^{no}
Strap wings 	vg^{st}
Vestigial wings 	vg

Example: ABO Blood Group in Humans

- Controlled by gene **I** with three alleles: **I^A**, **I^B**, **i**.
- I^A** and **I^B** are **codominant**, while **i** is **recessive**.
- Possible combinations and phenotypes:

ABO Blood Group System

Genotype	Phenotype
I^AI^A, I^Ai	Group A
I^BI^B, I^Bi	Group B
I^AI^B	Group AB
ii	Group O

Significance:

- Explains blood transfusion compatibility.
- Shows how one gene can exist in several forms, producing more variety than simple dominance.

2. Pleiotropy**From Greek:**

- pleion** = “more”
- tropos**=“change”
→ meaning “one gene, many effects.”
- Definition:** When a **single gene controls multiple traits** (characters), it is called pleiotropy.
- Happens because the protein/enzyme coded by the gene may be involved in **different metabolic pathways**.
- The ratio is 1:2 instead of 3:1 because of the death of recessive homozygote.

Examples

- Sickle-cell anemia (HBs gene mutation):**

- Change in hemoglobin β -chain.
- Leads to abnormal RBC shape (sickle-like).
- Causes anemia, reduced oxygen transport, and malaria resistance.

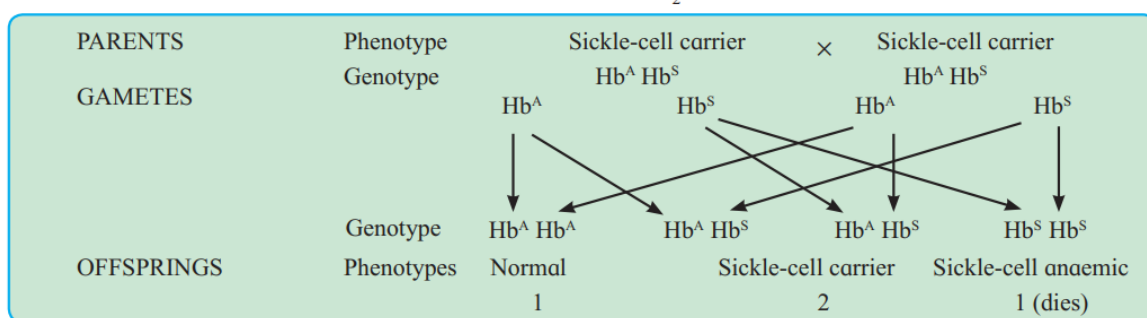
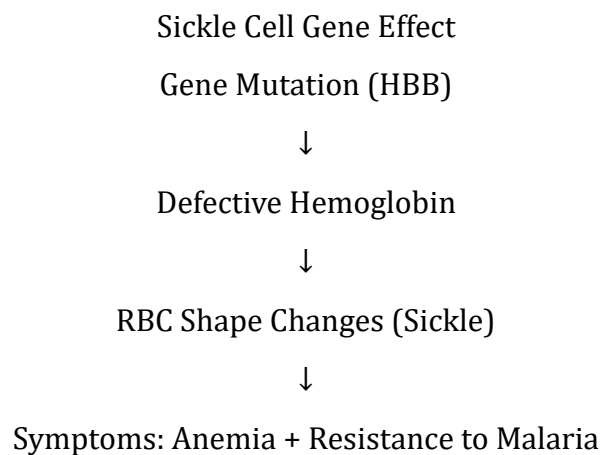


Fig. 3.4 : Representation of Pleiotropy

3.6 Chromosomal Theory of Inheritance :

- Due to advancements in microscopy, scientists observed cell division and the structure of chromosomes.
- **Proposed by:** Sutton & Boveri (1902–1903).
- **Key Idea:** Genes are located on chromosomes, and their **behaviour during meiosis** explains inheritance.

Main Postulates

1. Chromosomes are present in pairs in somatic cells
2. During gamete formation, homologous chromosomes pair, segregate and assort independently.
3. Thus, each gamete contains only one chromosome from a pair.

4. The nucleus of gametes contains chromosomes, which carry all hereditary traits.
5. The fusion of a haploid male gamete and a haploid female gamete restores the diploid number of chromosomes of the species.

3.7 Chromosomes :

“Chromosomes are filamentous structures present in the eukaryotic nucleus, composed of DNA and proteins, and they carry the genetic information in the form of genes.”

coined by **W. Waldeyer (1888)**

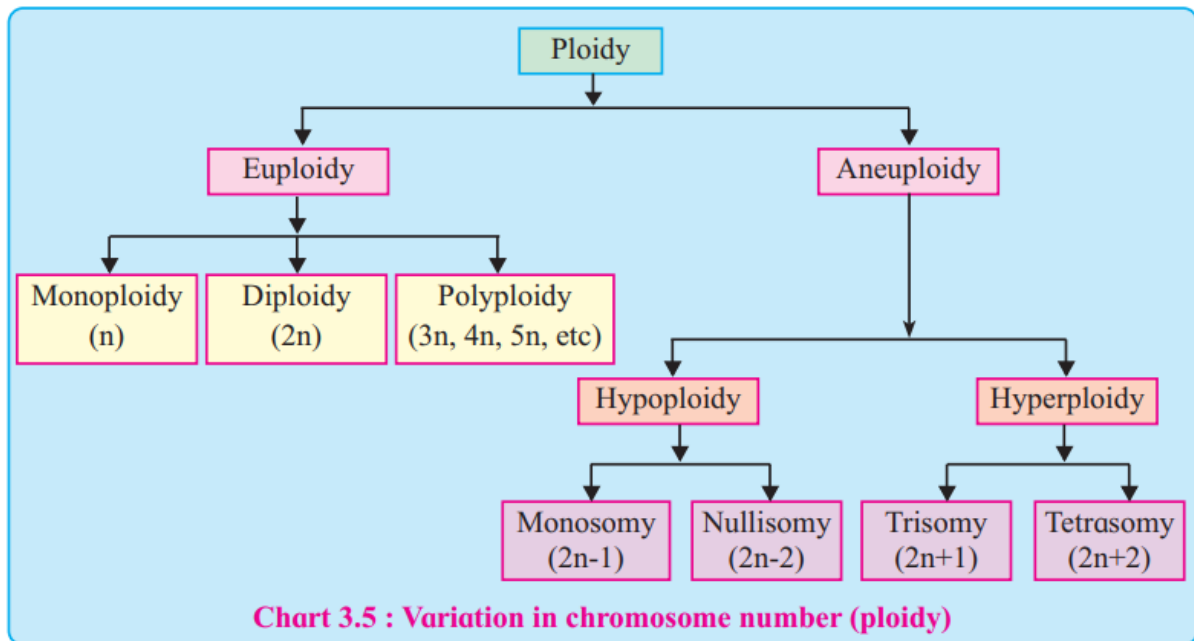
1 Properties:

- Size: 0.1 to 33 micrometres in length and 0.2 to 2 micrometers in thickness.
- Chromosomes are visible during cell division.
- Capable of self-replication.
- Plays a vital role in heredity, mutation, variation, and evolutionary development of eukaryotic species.

2 **Function:** Chromosomes mainly act as carriers of heredity.

3 **Number of chromosomes**

- specific and constant for a particular species
- **Ploidy:** degree of repetition of the primary basic number of chromosomes (i.e. 'x')
- **Euploidy:** When the chromosome number in a cell is **an exact multiple of the primary basic number**, then it is called **euploidy**



- **Triploid (3n):** Banana, seedless watermelon
- **Tetraploid (4n):** Potato, cotton, groundnut
- **Pentaploid (5n):** Dog rose (*Rosa canina*)
- **Hexaploid (6n):** Wheat, kiwi, oat

4 Structure of chromosome :

Chromosomes are **best visible** under a microscope when the cell is at the **metaphase stage**. At this stage, chromosomes are **highly condensed**.

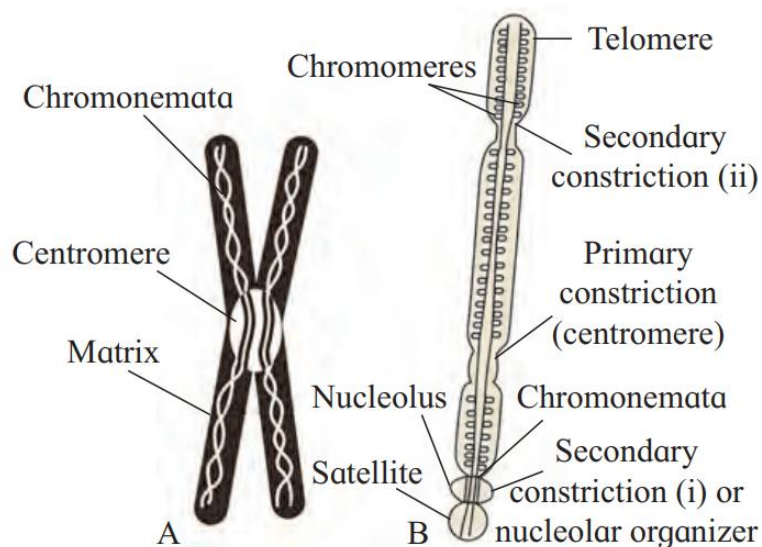
1. Primary Constriction (Centromere)

- Each chromosome has a **primary constriction** called the **centromere**.
- At the centromere lies a **disc-shaped plate** called the **kinetochore**.
- **Function:** The **spindle fibres attach** to the kinetochore during cell division, ensuring proper movement of chromosomes.

2. Secondary Constriction

- Some chromosomes possess **one or two additional constrictions** apart from the centromere.
- These are called **secondary constrictions**.
- **Secondary Constriction I:**
 - Involved in the organization of the **nucleolus during interphase**.
- **Secondary Constriction II:**
 - Found in very few chromosomes.

- Bears a small fragment called the **satellite body (SAT body)**.



A : Parts of chromosomes B : Showing secondary constrictions and details

Fig. 3.6 : Structure of Chromosome

3. Chromatid and Chromonema

- Each chromosome consists of **two chromatids** (sister chromatids).
- Each chromatid contains a **long, slender, unbranched, highly coiled DNA thread** called the **chromonema**, which extends throughout its length.
- Each chromatid contains **one double-stranded DNA molecule** that runs from **one end of the chromosome to the other**.

Types of Chromosomes (Based on Position of Centromere)

(Position of centromere, Length of arm, shape of chromosome)

1. Metacentric Chromosome

- Centromere is present **in the middle**.
- Both arms of the chromosome are **equal in length**.
- Appears **V-shaped** during anaphase.

2. Submetacentric Chromosome

- Centromere is **slightly away from the middle**.

- Produces **one short arm and one long arm**.
- Appears **L-shaped** during anaphase.

3. Acrocentric Chromosome

- Centromere is **near one end**.
- Results in a **very short arm and a very long arm**.
- Appears **J-shaped** during anaphase.

4. Telocentric Chromosome

- Centromere is **at the terminal end**.
- Chromosome has only **one arm**.
- Appears **I-shaped** during anaphase.

Table: Types of Chromosomes (Based on Position of Centromere)

Type of Chromosome	Position of Centromere	Length of Arms	Shape of Chromosome
Metacentric	In the middle	Two arms of equal length	V-shaped
Submetacentric	Slightly away from middle	One arm longer and one shorter	L-shaped
Acrocentric	Near one end	One arm very long , one very short	J-shaped
Telocentric	At the terminal end	Chromosome has only one arm	I-shaped

Telomeres

- The **ends of chromosomes (chromatids)** are called **telomeres**.
- Function: Protect chromosome ends and prevent them from fusing with one another.

Sex Chromosomes (Allosomes)

Definition

- Chromosomes responsible for **determination of sex** are called **sex chromosomes** (allosomes).
- In humans and other mammals, these are the **X and Y chromosomes**.

X Chromosome

- **Shape & Size:** Straight, rod-like, **longer** than Y chromosome.
- **Type:** Metacentric.
- **Chromatin Content:**
 - Large amount of **euchromatin** (extended region, metabolically active).
 - Small amount of **heterochromatin** (condensed region).
- **Activity:** **Genetically more active** due to higher euchromatin content.

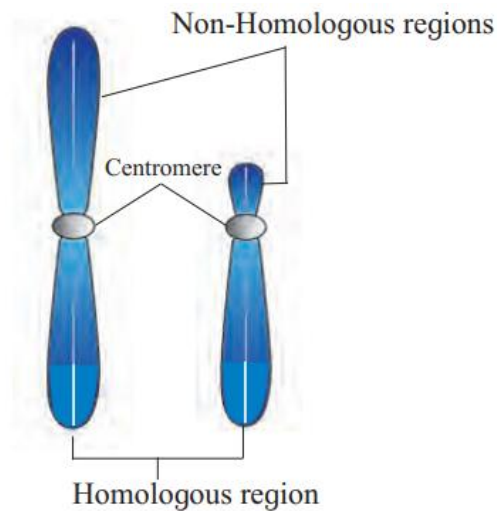
Y Chromosome

- **Shape & Size:** **Shorter** than X chromosome.
- **Type:** Acrocentric.
- **Chromatin Content:**
 - Small amount of **euchromatin** (less active).
 - Large amount of **heterochromatin** (inert region).
- **Activity:** **Genetically less active or inert** compared to X chromosome.

Homologous vs. Non-homologous Regions

- **Homologous regions:** Present on both X and Y; contain **similar genes** (e.g., pseudoautosomal regions). Crossing over occurs at homologous regions of both chromosomes

- **Non-homologous regions:** Present only on one chromosome; contain **different genes** (responsible for sexual dimorphism). X chromosome has larger Non homologous Region than Y.



**Fig. 3.7 : Structure of X and Y chromosomes
(in humans)**

The difference between X and Y chromosome

Feature	X Chromosome	Y Chromosome
Size	Larger, longer	Smaller, shorter
Centromere position	Metacentric	Acrocentric
Chromatin content	Large amount of euchromatin (active)	Large amount of heterochromatin (inert)
Genetic activity	Genetically more active	Genetically less active / partially inert
Non-homologous regions	Longer , contains more genes (X-linked genes)	Shorter , contains fewer genes (Y-linked genes, e.g., SRY)
Function	Carries many essential genes, including non-sex traits	Carries male-determining genes and few others

- Crossing over occurs **only between homologous regions** of the X and Y chromosomes (pseudoautosomal regions).
- This ensures proper pairing and segregation of sex chromosomes during meiosis.

Non-homologous Regions

- **X chromosome non-homologous region:**
 - **Longer** than that of Y.
 - Contains **more genes**, many of which are **X-linked genes**.
- **Y chromosome non-homologous region:**
 - **Shorter** than that of X.
 - Contains fewer genes, many of which are **Y-linked genes** (e.g., SRY gene controlling male sex determination).

Gene Distribution

- **X-linked genes** → located on the non-homologous region of the **X chromosome**.
- **Y-linked genes** → located on the non-homologous region of the **Y chromosome**.

3.8 Linkage and crossing over:

Linkage

1. Definition

- **Linkage** is the tendency of **two or more genes located on the same chromosome** to be **inherited together** during cell division.
- Genes that are located close to each other on a chromosome are called **linked genes**.

2. Discovery

- Discovered in **plants** by **Bateson and Punnett** (Sweet pea experiments).
- Discovered in **animals** by **T. H. Morgan** (*Drosophila* studies).

3. Concept

- Since **chromosomes are carriers of heredity**, genes present on the same chromosome do not assort independently.
- Instead, they **tend to be transmitted together** from parent to offspring.

4. Types of Linkage

I. Complete Linkage

- **Definition:** When linked genes are **very close together** on a chromosome and **do not separate** (no crossing over).
 - Only **parental traits** are inherited.
 - No new combinations appear in offspring.
- **Example:** Complete linkage in the **X chromosome of *Drosophila* males**.

II. Incomplete Linkage

- **Definition:** When linked genes are located **farther apart** on the same chromosome and may **separate due to crossing over**.
 - **New gene combinations** (recombinants) appear in the offspring.
 - Along with **parental traits**, new traits are also seen.
- **Example:** In **Zea mays (maize)**, the genes for **colour and shape of grain** show incomplete linkage.

Feature	Complete Linkage	Incomplete Linkage
Definition	Genes located very close together on the same chromosome, so no crossing over occurs.	Genes located farther apart on the same chromosome, so crossing over may occur .
Inheritance pattern	Linked genes are always inherited together .	Linked genes are usually inherited together , but crossing over produces recombinants.
Result in offspring	Only parental traits appear.	Both parental and new (recombinant) traits appear.
Recombination	Absent (no new combinations formed).	Present (new combinations formed due to crossing over).
Strength of linkage	Genes are completely/strongly linked .	Genes are incompletely/weakly linked .
Example	X chromosome of <i>Drosophila</i> males (showing complete linkage).	<i>Zea mays</i> (maize) – genes for colour and shape of grain show incomplete linkage.

5. Significance of Linkage

- Helps in understanding **gene arrangement on chromosomes**.
- Provides evidence for **the chromosomal theory of inheritance**.
- Useful in **genetic mapping** (locating genes on chromosomes).

6. Linkage Groups

- **Definition:** All the linked genes present in a particular chromosome together form a **linkage group**.
- **Rule:** The number of linkage groups in a species = **haploid number of chromosomes**.

Examples

- *Drosophila melanogaster* → **4 linkage groups** (corresponds to 4 pairs of chromosomes).
- Garden pea (*Pisum sativum*) → **7 linkage groups** (corresponds to 7 pairs of chromosomes).

7. Sex Linkage (Sex-linked Inheritance)

- **Definition:** The inheritance of genes located on **X or Y chromosomes** from parents to offspring.
- **Types:**
 - **X-linked**
 - **Y-linked**
 - **XY-linked**

Kinds of Sex Linkage

a. Complete Sex Linkage

- Genes are located on the **non-homologous regions** of X and Y chromosomes.
- **Crossing over does not occur**, so genes are inherited **together**.
- **Examples:**
 - **X-linked traits:** Haemophilia, Red-green colour blindness, Myopia, Ichthyosis.
 - **Y-linked traits:** Hypertrichosis, H-Y antigen gene.

b. Incomplete Sex Linkage

- Genes are located on the **homologous regions** of X and Y chromosomes.
- **Crossing over occurs**, so genes **do not inherit together**.
- **Examples:** Total colour blindness, Nephritis, Retinitis pigmentosa.

2. Crossing Over

- **Definition:** Process that produces **new gene combinations (recombinants)** by exchange of corresponding segments between **non-sister chromatids of homologous chromosomes**.
- **Stage:** Occurs during **pachytene of prophase I of meiosis**.
- **Coined by:** The Term *crossing over* was given by **T. H. Morgan**.

Steps of Crossing Over

1. **Synapsis** – Pairing of homologous chromosomes.
2. **Tetrad formation** – Four chromatids come together.
3. **Crossing over** – Exchange of chromatid segments between non-sister chromatids.
4. **Terminalization** – Chiasmata move towards the chromosome ends.

Significance

- Crossing over is **universal** (except in *Drosophila* males).
- Creates **genetic recombination** → increases **variation**.
- Variation is essential for **natural selection and evolution**.

Morgan's Experiments: Linkage and Crossing Over

1. Why Morgan chose *Drosophila melanogaster* (fruit fly)?

- **Easy to culture** in laboratory.
- **Short life span** (~2 weeks).
- **High rate of reproduction**.
- **Large number of visible traits** (eye colour, body colour, wing type).
- **Few chromosomes** ($2n = 8$), making genetic studies easier.

2. Experimental Design

- Morgan studied **dihybrid crosses** in *Drosophila*, similar to Mendel's pea plant experiments.
- Aim: To study **sex-linked genes** and their inheritance.

Example Cross:

- **Parental cross (P):**
 - Yellow-bodied, white-eyed female (mutants) × Brown-bodied, red-eyed male (wild type).
- **F1 Generation:**

- All flies showed **wild type phenotype** (brown body, red eyes).
- Indicates dominance of wild traits.
- **F₂ Generation (F₁ intercross):**
 - Did not show Mendel's expected **9:3:3:1 ratio**.
 - Instead, observed a higher proportion of **parental combinations** and a smaller number of **new (recombinant) combinations**.

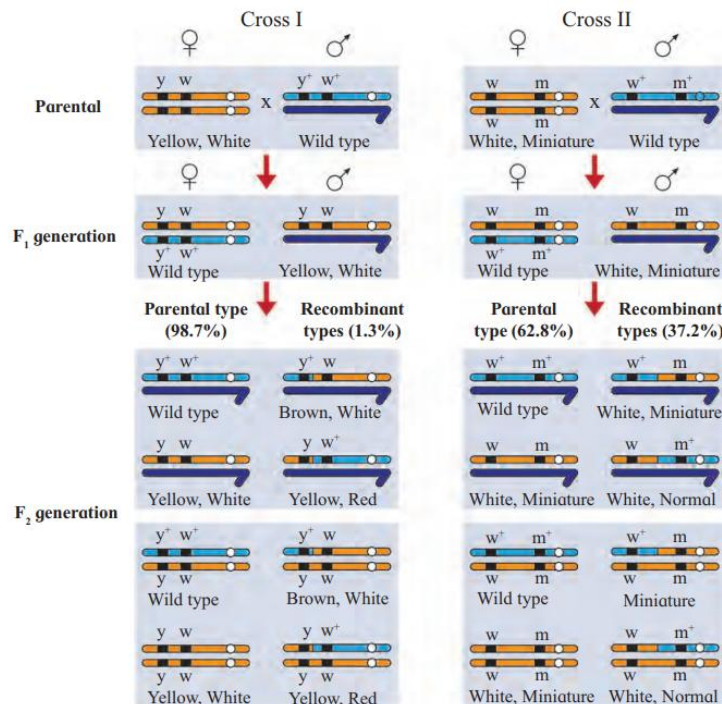


Fig : 3.8 : Linkage and crossing over

Cross I

- Genes: y (yellow body colour) and w (white eye colour).
- Very closely linked on the same chromosome.
- **Results:**
 - Parental types: 98.7% (almost no recombination).
 - Recombinants: 1.3% (rare crossing over).
- Indicates complete or tight linkage, where genes are almost always inherited together.

Cross II

- Genes: w (white eye) and m (miniature wing).
- Located farther apart on the same chromosome.
- **Results:**

- Parental types: 62.8%
- Recombinants: 37.2%
- Indicates incomplete linkage, as crossing over occurs more frequently.

Key Concepts

- Linkage: Genes on the same chromosome tend to be inherited together.
- Crossing Over: Exchange of segments between non-sister chromatids during prophase I (pachytene stage) of meiosis, producing recombinant gametes.
- Genes in close proximity show fewer recombinants (Cross I), while distant genes show higher recombination frequencies (Cross II).

Observations

- Genes for body color and eye color were linked.
- Parental types dominated over recombinants.
- Limited recombinants arose due to crossing over.

Conclusion

- Linkage ensures inheritance of parental combinations.
- Crossing over between homologous chromosomes produces recombinants, showing incomplete linkage.
- Morgan's experiments provided clear evidence of linkage and crossing over.

Significance of Morgan's Experiments

- Offered cytological proof of the chromosomal theory of inheritance.
- Led to the development of linkage maps using recombination frequency.
- Demonstrated that linkage modifies Mendel's law of independent assortment, highlighting exceptions.

3.9 Autosomal Inheritance

Transmission of body characters other than the sex linked traits from parents to their offspring through autosomes is called autosomal inheritance.

1. Definition

- **Autosomal traits** are characters determined by genes located on the **autosomes** (non-sex chromosomes).
- These traits can be influenced either by **dominant** or **recessive alleles**.
- Since autosomes are present in **both sexes equally**, these traits show **equal inheritance in males and females**.

2. Categories of Autosomal Traits

A. Autosomal Dominant Traits

- A single **dominant allele** is sufficient to express the trait.
- Individuals with **homozygous dominant (AA)** or **heterozygous (Aa)** genotype exhibit the trait.
- Example traits/disorders:
 - **Widow's peak**
 - **Huntington's disease**

B. Autosomal Recessive Traits

- Trait is expressed only when **both alleles are recessive (aa)**.
- **Carriers (Aa)** do not express the trait but can pass it to offspring.
- Example disorders:
 - **Phenylketonuria (PKU)**
 - **Cystic fibrosis**
 - **Sickle-cell anaemia**

3. Example: Widow's Peak

- **Description:** A prominent "V"-shaped hairline in the middle of the forehead.
- **Genetic basis:** Controlled by an **autosomal dominant allele (W)**.
- **Genotypes:**
 - **WW or Ww** → Widow's peak present.
 - **ww** → Straight hairline (no widow's peak).

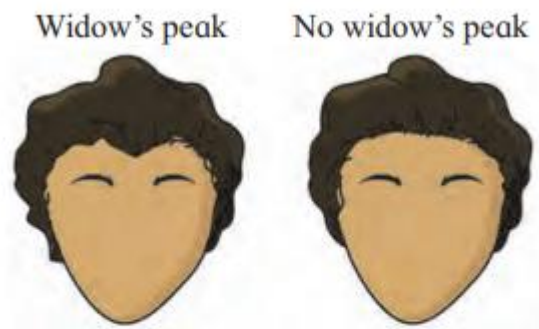


Fig. 3.9 : Widow's peak and straight hair line

- **Inheritance pattern:**
 - Appears in **both sexes equally**.
 - Can be inherited from **either parent**, as the gene is on an autosome.

2. Phenylketonuria (PKU)

- **Type:** Autosomal recessive disorder (inborn metabolic disease).
- **Cause:** Mutation in **recessive autosomal genes** → enzyme **phenylalanine hydroxylase** not produced.
- **Normal role of enzyme:** Converts amino acid **phenylalanine** → **tyrosine**.
- **Defect:**
 - Phenylalanine not converted.
 - Accumulation of **phenylalanine and derivatives** in blood and cerebrospinal fluid (CSF).
- **Effects:**
 - Severe damage to brain development.
 - Mental retardation.
 - **Excretion of excess phenylalanine in urine** → hence called *Phenylketonuria*.
- **Inheritance:** Appears in both sexes **equally**, but only in **homozygous recessive** individuals.

Key Point

- **Autosomal recessive traits** (like PKU) → appear in both sexes **equally**, often **skip generations**.
- **X-linked recessive traits** (like Colour blindness) → more frequent in **males**, rare in females.

3.10 Sex-Linked Inheritance

1. Definition

- **Sex-linked genes** → Genes located on the **non-homologous region of sex chromosomes (X or Y)**.
- **Sex-linked traits** → Traits determined by these genes.
- **Sex-linked inheritance** → Transmission of sex-linked genes from **parents to offspring**.

2. Types of Sex-Linked Genes

- **X-linked genes:** Located on the **non-homologous region of X chromosome**.
- **Y-linked genes:** Located on the **non-homologous region of Y chromosome**.

3. X-linked Genes

- Present on **X chromosome** but **absent on Y chromosome**.

- **Inheritance pattern differs between sexes:**

In Females (XX):

- Two X chromosomes present.
- Expression of a recessive X-linked trait requires **two recessive alleles** (X^cX^c).
- If only one recessive allele is present (XCX^c), its effect is **masked by the dominant allele** on the other X chromosome.
- Such females are **carriers** → phenotypically normal but can transmit the disorder.

Examples:

1. Colour Blindness

- **Type: X-linked recessive disorder.**
- **Cause:** Recessive gene (X^c) prevents formation of **cone cells** in retina → inability to distinguish **red and green colours** (appear grey).
- **Inheritance pattern:**
 - **Males (XY):** Only one X chromosome, so a single recessive gene (X^cY) makes them colour blind.
 - **Females (XX):** Need **two recessive alleles** (X^cX^c) to be colour blind.
 - **Carrier females (X^CX^c):** Normal vision, but can pass gene to offspring.
- **Frequency:** More common in males than females (because males are hemizygous for X chromosome).

Sex	Normal Vision	Carrier	Colour Blind
Male	X^CY	–	X^cY
Female	X^CX^C	X^CX^c	X^cX^c

Example: Inheritance through Marriage

Cross 1: Colour blind male (X^cY) × Normal female ($XCXC$)

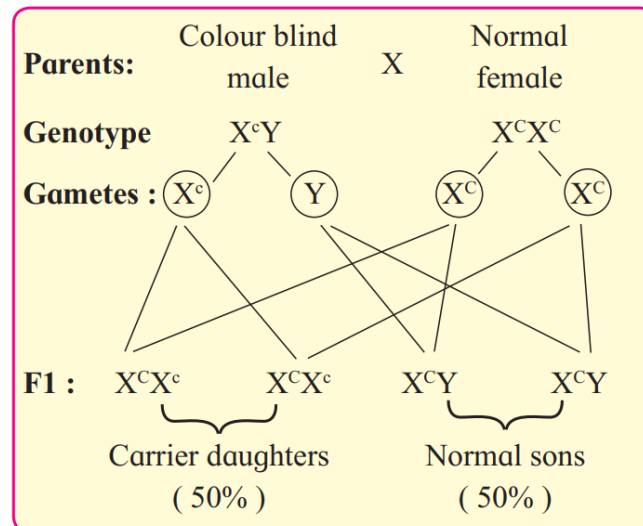


Fig. 3.10 : Sex linked inheritance (colour blindness)

- **F1 Progeny:**
 - **Sons:** X^CY → Normal vision.
 - **Daughters:** X^CX^c → Normal vision but **carriers**.

Thus, disorder **skips generations** when carried by females.

In Males (XY):

- Only **one X chromosome** is present.
- If the X chromosome carries a recessive allele (X^cY), the trait is **expressed**, because there is **no corresponding dominant allele on Y chromosome**.
- Hence, **X-linked traits are more frequent in males** than in females.

Case 2: carrier female (daughter) and normal male

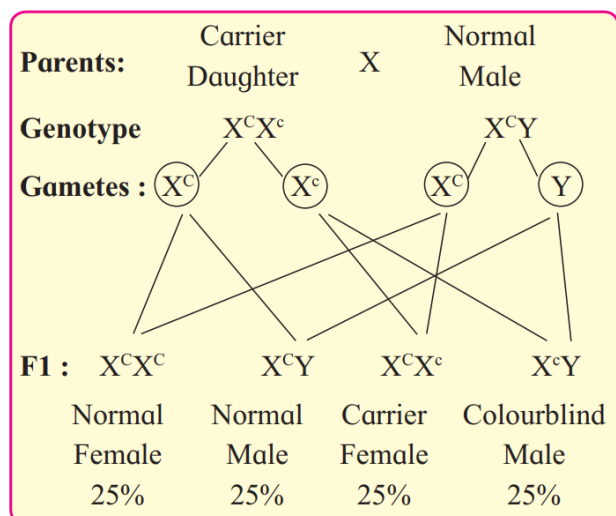


Fig. 3.11 : Sex linked inheritance (colour blindness)

From above example:

- X linked recessive gene for colour blindness is inherited from colourblind father to his grandson through his daughter.
- This type of inheritance is called as criss-cross inheritance.

Haemophilia (Bleeder's Disease)**Definition**

- An **X-linked recessive disorder** where blood **fails to clot normally** or clots very slowly.
- Leads to prolonged bleeding even from **minor injuries**.

Cause

- Deficiency of blood clotting factors:
 - **Factor VIII** → Haemophilia A (classical type)
 - **Factor IX** → Haemophilia B (Christmas disease)
- Caused by recessive gene located on the **non-homologous region of X chromosome**.

Inheritance

- **X-linked recessive:**
 - Males (XY) are more frequently affected → because they have only one X chromosome, no corresponding allele on Y to mask the defect.
 - Females (XX) usually **carriers** if they inherit one defective gene; only suffer disease if **both X chromosomes** carry the recessive allele (very rare).

Symptoms

- Excessive or continuous bleeding from small cuts/injuries
- Spontaneous internal bleeding (into joints, muscles, brain)
- Delayed wound healing
- Bruising easily

Other Names

- **"Bleeder's disease"** — due to uncontrolled bleeding tendency

Key Points

- Dominant allele = normal clotting
- Recessive allele = defective clotting → haemophilia
- More common in **males**, rare in females
- Historically known as the **"royal disease"** (found in Queen Victoria's descendants)

Case 1: Haemophilic male and normal female

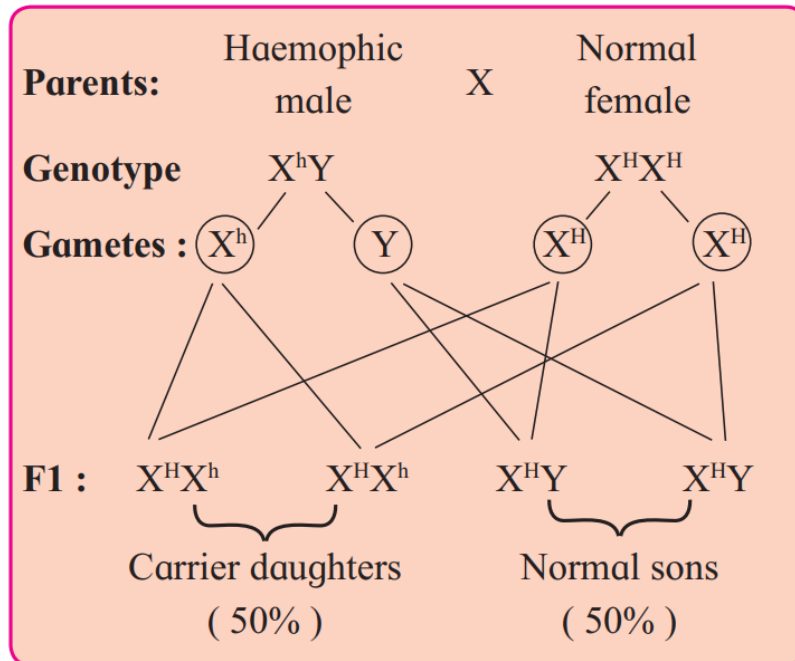


Fig. 3.12 : Sex linked inheritance (Haemophilia)

Case 2. Carrier female (daughter) and normal male:

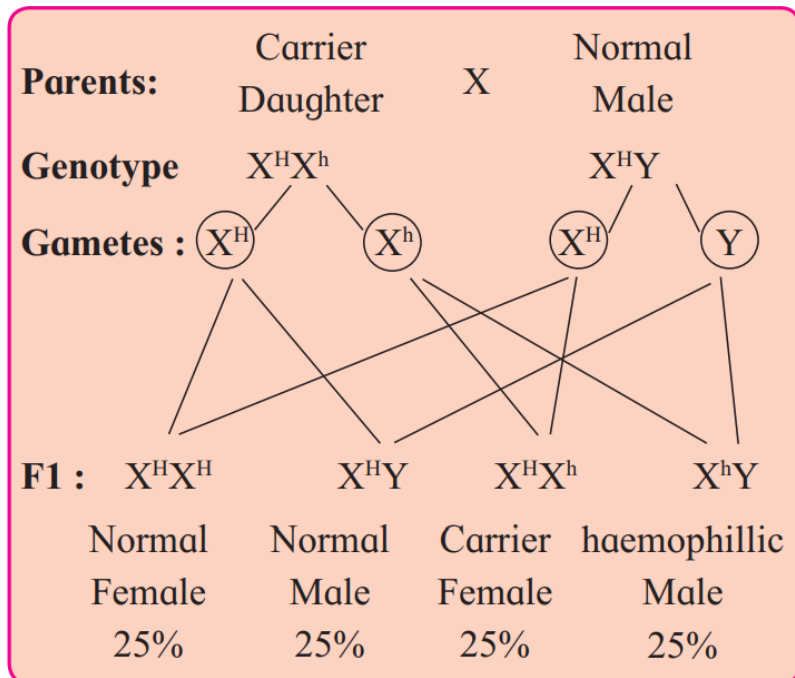


Fig. 3.13 : Sex linked inheritance (Haemophilia)

4. Examples of X-linked Traits

- Haemophilia
- Red-green colour blindness
- Night blindness
- Myopia (near-sightedness)
- Muscular dystrophy
-

Comparative chart of Autosomal and Sex Inheritance

Feature	Autosomal Inheritance	Sex-Linked Inheritance
Chromosomes involved	Genes located on autosomes (22 pairs of non-sex chromosomes)	Genes located on the sex chromosomes (X or Y)
Traits controlled	Traits other than sex determination (body traits)	Traits determined by genes present on X or Y chromosomes
Occurrence in sexes	Traits appear in both males and females equally	X-linked traits: more common in males (as they have only one X); Y-linked traits: only in males
Expression of recessive traits	Both sexes need homozygous recessive condition for trait to express (e.g., PKU, cystic fibrosis)	In females , two recessive X-linked genes needed; in males , one recessive X gene is enough (hemizygous condition)
Carriers	No carrier condition , because autosomal traits express equally in both sexes	Carrier females possible (heterozygous with one defective X gene); males cannot be carriers for X-linked traits
Transmission	Trait passes from parents to offspring through autosomes	Trait passes through sex chromosomes (X or Y)
Examples (dominant)	Widow's peak, Huntington's disease	– (X-linked dominant disorders are rare, e.g., vitamin D-resistant rickets)
Examples (recessive)	PKU, cystic fibrosis, sickle cell anaemia	X-linked: haemophilia, colour blindness, night blindness, muscular dystrophy; Y-linked: hypertrichosis (hairy pinna of ear)
Inheritance pattern	Equal chance of inheritance in both sexes	- X-linked recessive: mostly males affected; females may be carriers - Y-linked: only father → son transmission

3.11 Sex determination:

The mechanism by which sex is established is termed as sex determination.

- In some species, both male and female reproductive organs are present in same organism.
- They are bisexual or hermaphrodite or monoecious.
- species in which the organism has either male or female reproductive organs, is said to be dioecious or unisexual.
- Humans are dioecious.

Henking in 1891, studied the spermatogenesis in Squash bug (*Anasa tristis*) and found that 50 % chromosome receives unpaired chromosome and 50% don't.

He called it as x-body but later other scientist named it as X chromosome.

a. Sex Determination in human beings : (XX-XY Type)

- In human the mechanism of sex determination is XX-XY type.
- In each nucleus of somatic cell there are 46 chromosomes or 23 pairs of chromosomes.
- Out of 46 chromosome 44 are autosomes which have body characters and 2 are for the sex determination of individual.
- Means , 22 pairs are autosomes and one pair of sex chromosomes.

Individual	Genotype	Gametes Produced (meiosis product) (oogenesis / Spermatogenesis)
Female	44 Autosomes + XX	All gametes contain 22 autosomes + X
Male	44 Autosomes + XY	22 autosomes + X 22 autosomes + Y

Fertilisation Outcome:

Female Gamete (Egg)	Male Gamete (Sperm)	Resulting Zygote	Sex of Offspring	Probability
22A + X	22A + X	44A + XX	Female	50%
22A + X	22A + Y	44A + XY	Male	50%

Hence, the father is responsible for determining the sex of the child, not the mother.

Due to a lack of knowledge, women are often blamed for giving birth to a female child.

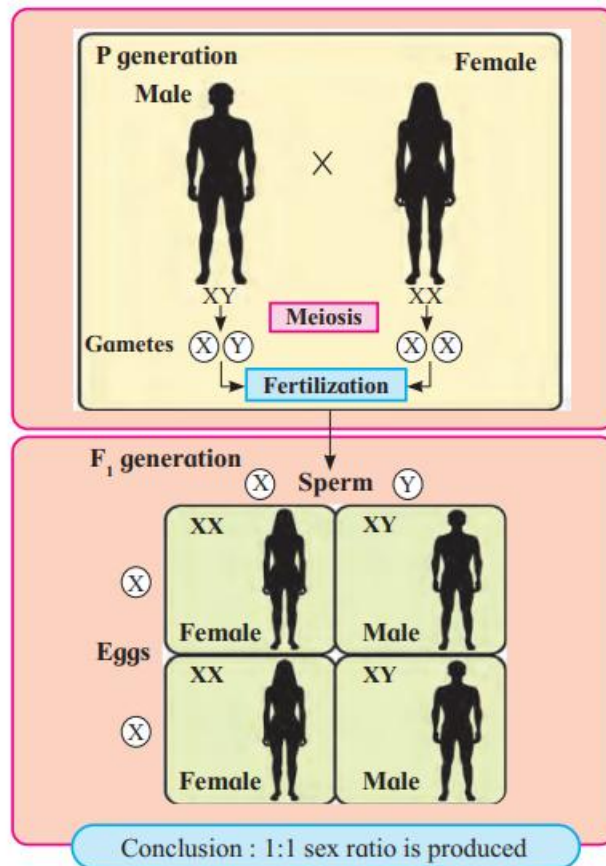


Fig. 3.14 : Sex determination in human beings

b. Sex Determination in Birds: ZW-ZZ type

In birds, the chromosomal mechanism of sex determination is the ZW-ZZ type.

Here, females are heterogametic and produce two types of eggs; 50% eggs carry the Z chromosome, while 50% eggs carry the W- chromosome.

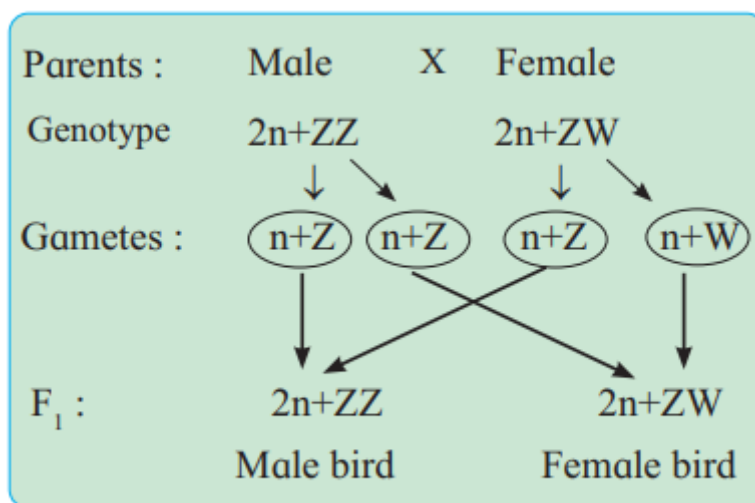


Fig. 3.15 : Sex determination in birds

Males are homogametic and produce one type of sperm. Each sperm carries a Z chromosome.

Thus, the sex of an individual depends on the kind of egg (ova) fertilised by the sperm.

c. Sex Determination in honey bees : haplo-diploid type

- In this case, the sex of an individual is determined by the number of sets of chromosomes received.
- Females are diploid ($2n=32$) and males are haploid ($n=16$).
- The female produces haploid eggs ($n=16$) by meiosis, and the male produces haploid sperm ($n=16$) by mitosis.
- If the egg is fertilized by sperm, the zygote develops into a diploid female ($2n=32$) (queen and worker).
- Unfertilised egg develops into haploid male ($n=16$) (Drone) by way of parthenogenesis

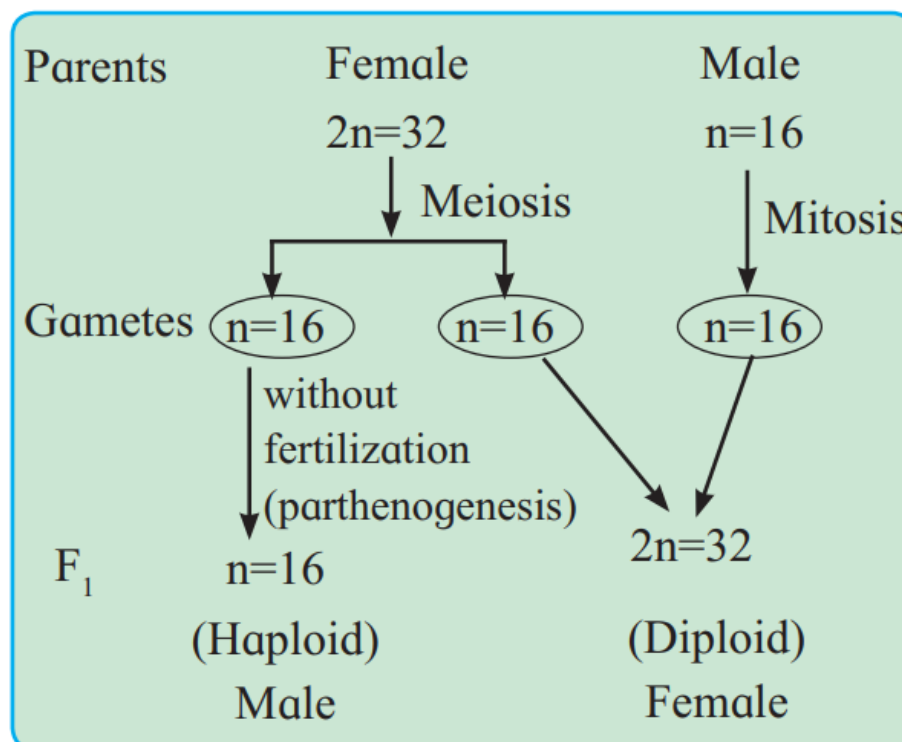


Fig. 3.16 : Sex determination in honey bee

- A worker or a Queen is determined by the food they get.
- Diploid larvae, which get royal jelly as food, develop into queens (fertile females) .
- Other develops into workers (sterile females).

3.12 Genetic Disorders :

Genetic Disorders are broadly grouped into two categories as, Mendelian disorders and chromosomal disorders.

Category	Cause	Examples
Mendelian Disorders	Alteration or mutation in a single gene	Thalassemia, Sickle-cell anaemia, Colour blindness, Haemophilia, Phenylketonuria
Chromosomal Disorders	Absence, excess, or abnormal arrangement of chromosomes	Down's syndrome, Turner's syndrome, Klinefelter's syndrome

Mendelian Disorders

1. Thalassemia: Mendelian disorder

- Thalassemia is an autosomal, inherited recessive disease
- Haemoglobin molecule is made of four polypeptide chains- 2 alpha (D) and 2 beta (E) chains.
- The chains are controlled by two closely linked genes (HBA1 and HBA2) on chromosome 16 while beta chain is controlled by a single gene (HBB) on chromosome 11.
- Depending upon which chain of haemoglobin is affected, thalassemia is classified as alpha-thalassemia and betathalassemia.
- The disorder result in abnormal synthesis of haemoglobin.
- Symptoms :
 - Anaemia,
 - Pale yellow skin,
 - Change in size and shape of rbcs,
 - Slow growth and development,
 - Dark urine, etc.

Chromosomal Disorders

2. Down's syndrome: (21st trisomy)

- Named after the physician John Langdon Down who first described this autosomal chromosomal disorder in 1866.
- Caused due to an extra copy of chromosome number 21st.
- Have 47 chromosomes instead of the normal number 46
- 21st trisomy occurs due to non-disjunction or failure of separation of chromosomes. (autosomes) during gamete formation.
- Mothers who are over 45 years old have higher chances of non-disjunction.
- Symptoms:
 - mild or moderate mental retardation
 - skeletal development is poor



Fig. 3.17 : Down's Syndrome

- Distinct facial features like small head, ears and mouth, face is typically flat and rounded with flat nose, open mouth and protruding tongue, eyes slant up and out with internal epicanthal folds,
- flat hands and stubby fingers and palm is broad with single palmer crease.

Down Syndrome - Trisomy 21

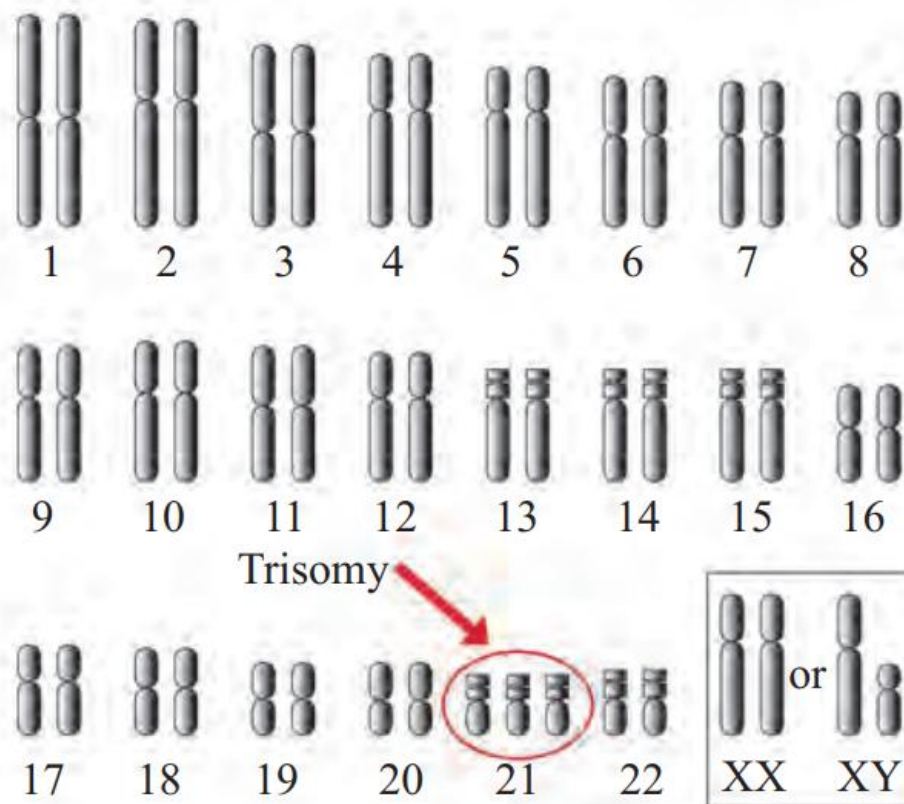


Fig. 3.18 : Karyotype of Down's syndrome

3. Turner's Syndrome : (X monosomy / XO females)

caused due to non-disjunction of chromosome during formation of egg.

Turner's syndrome has 44 autosomes with XO.

They are phenotypically female.

Symptoms:

- ✓ Short stature (height) and
- ✓ Webbed neck,
- ✓ Lower posterior hair line,
- ✓ Broad shield-shaped chest,
- ✓ Poorly developed ovaries and breasts and
- ✓ Low intelligence.

4. Klinefelter's syndrome (XXY males) :

- It is caused by an extra X chromosome in males.
- Thus, the genotype of individuals is 44 + XXY.
- They are described as feminised males.
- An extra chromosome is a result of non-disjunction of the X chromosome during meiosis in the formation of an ovum.

Symptoms:

- ✓ Tall with long arms
- ✓ Voice pitch is harsh
- ✓ Feminine development (development of breast i.e. Gynaecomastia)
- ✓ Underdeveloped testis
- ✓ No spermatogenesis, hence sterile

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Some more treasure:

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