CHAPTER → 5 PRINCIPLES OF INHERITANCE AND VARIATION

* **Genetics** → Deals with inheritance and Variation of characters from parents to offspring.
* **Inheritance** → process by which, characters are passed from parent to progeny
* **Variation** → Degree by which progeny differs from their parents.

Humans Knew from 8000 - 1000 B.C.
↳ Cause of variation was hidden in Sexual reproduction → Exploited Variation → Selectively Breed, (Desirable characters) → Artificial selection and domestication
↓
Sahiwal Cows in Punjab.

* MENDEL'S LAW OF INHERITANCE :→ 1856 - 1863 (Mid 19th Century) → 7 Years

↳ Gregor Mendel → Austrian Monk
↓
Conducted Hybridisation experiments on garden pea Pisum Sativum.

* Why Mendel Got Success ?

① Application of mathematical and Statistical logics.
② Large Sampling Size.
③ Investigated on Opposite traits (Tall, Dwarf .. Yellow Green)
④ Meticulous Records.
⑤ Conducted artificial / Cross pollination on several true breeding lines.

* Mendel Selected 14 true breeding pea plant Varieties as pairs, were similar except for one character.

* True-Breeding line :→

↓
Continuous Self pollination.
↓
Stable trait inheritance and expression for several generat.

* 7 pairs of Contrasting traits in Pea :→

Character	Dominant Trait	Recessive Trait
Plant Height	Tall	Dwarf
Flower Position	Axial	Terminal
Flower Colour	Violet	White
Seed Shape	Round	Wrinkled
Seed Colour	Yellow	Green
Pod Shape	Full	Constricted
Pod Colour	Green	Yellow

* INHERITANCE OF ONE GENE :

↓
By Mendel → Hybridisation experiment.

One character
Tall Pea Plant × Dwarf Pea Plant
↓
Seeds
(F1 generation, Filial progeny)

TT × tt (Monohybrid Cross)
↓
Tt Heterozygous tall
(F1 generation) →
All of these were tall.

→ F1 always resemble either of the parents and characters of other parent cannot be seen in F1 generation.

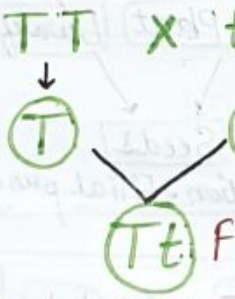
* Steps in Making a Cross in Pea :→

Emasculation → Bagging → Transfer of desired pollen → Rebagging.

* Definations :→

1. **Factors / Genes** → Something which is stably passed from parents to offspring.
2. **Unit of Inheritance** → Genes
3. **Basis of Heredity** → Inheritance.
4. **Allels** → Genes which code for contrasting pair of traits. i.e. Slightly diff. forms of same gene.
5. **Gene** contain information required to express particular trait in organism.
6. **Genotype** → Genetic Constitution of Character
7. **Phenotype** → Visibility of Character.
8. **Homozygous** → Same genes in a gamete. e.g. (TT).
9. **Heterozygous** → Diff genes in a gamete. e.g. (Tt)

* INHERITANCE OF ONE GENE / (ONE CHARACTER) MONOHYBRID :→



All Heterozygous Tall.

Selfing → Tt x Tt

Gametes →

	T	t
T	Tt	Tt
t	Tt	tt

} F2 Generation

Tall and dwarf were identical to parent type.

* PUNNET SQUARE :→

↳ British Geneticist → Reginald C. Punnett.

Graphical representation to calculate probability of all possible genotypes of offspring in a genetic cross

* Ratio's On Selfing :→

1. **Phenotypic Ratio** →

Tt Tt TT : tt

Tall : dwarf

3 : 1

2. **Genotypic Ratio** →

TT : Tt, Tt : tt

1 : 2 : 1

→ Contrasting traits did not show any blending at F_1 or F_2 stage.

Gametes are produced by meiosis, allels of parental pair segregate from each other and only one allel is transmitted to gamete

Segregation is Random process

50% chance of a gamete containing either allel, has been verified by results of crossings.

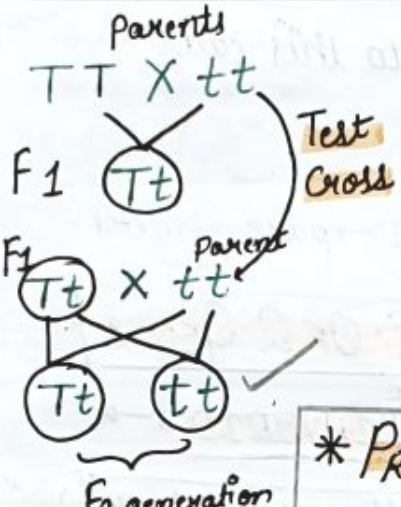
$$\left(\frac{1}{2}T+\frac{1}{2}t\right)^2=\left(\frac{1}{2}T+\frac{1}{2}t\right)\times\left(\frac{1}{2}T+\frac{1}{2}t\right)$$

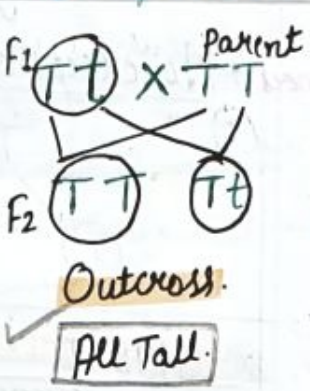
$$=\frac{1}{4}TT+\frac{1}{2}Tt+\frac{1}{4}tt$$

* **TEST CROSS** → Mating with recessive parent in order to know the genotype of plant.

* **OUT CROSS** → When F_1 progeny is crossed with dominant parents.

* **BACK CROSS** → Breeding of F_1 generation with one of its parents.





* PRINCIPLES / LAWS OF INHERITANCE →

First Law → Law Of Dominance

↳ Based on Observation of Monohybrid Cross.

1. Characters are controlled by discrete Units called Factors.
2. Factors occurs in pairs.
3. In dissimilar pair of factors, one is dominant and other recessive.

↳ **It Explains :→**

1. Expression of only one parental character in F_1 and expression of Both in F_2.

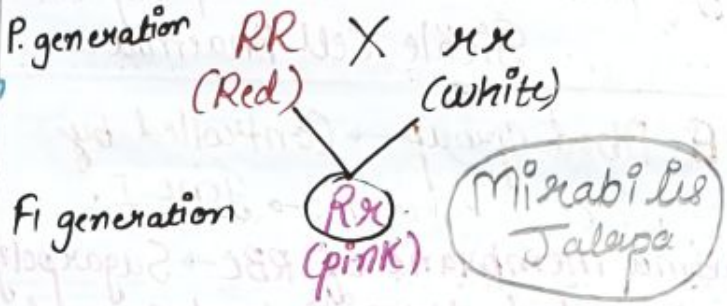
↳ **Exceptions to this Law :→**

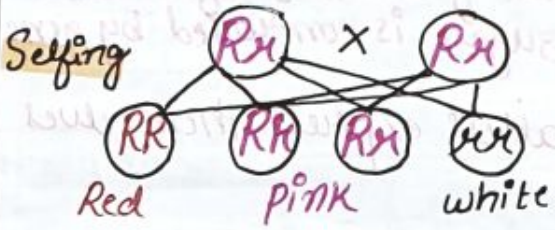
1. ~~Incomplete Dominance~~
2. ~~Co-dominance~~

① Incomplete Dominance :→

Incomplete Dominance forms an Intermediate trait.

Eg → Snapdragon or Antirrhinum sp.





Phenotypic Ratio → 1:2:1
Genotypic Ratio → 1:2:1

Ratio is Same.

Gene → Contains info to express a particular trait.
↳ e.g - Containing information for producing an enzyme.

Normal allel → Normal enzyme → To unmodified (functioning) allel → represents original phenotype → Dominant

Modified allel → Normal / less efficient → Kinda Unmodified
→ Non-functional → No enzyme
These will affect allele

(2) Co-Dominance :→

↳ In Co-dominance F1 generation resembles both parents.

* Examples → Human Blood Group (ABO)
Sickle Cell Anaemia.

ABO Blood Group → Controlled by gene I.

plasma membrane of RBC → Sugar polymers that protude from its surface and kind of sugar is controlled by gene.

↳ Both allels express themselves fully.

↳ A and B allels produce AB blood group.

* MUTIPLE ALLELS :→

↳ Eg → Human Blood Group:

↳ Each human has two allels for determination of Blood group, but population has 3 allels. A, B, O.

↳ Multiple allels are found when population study is made.

Allel from parent 1	Allel from parent 2	Genotype of offspring	Blood types of offspring
I^A	I^A	I^AI^A	A
I^A	I^B	I^AI^B	AB
I^A	i	I^Ai	A
I^B	I^A	I^AI^B	AB
I^B	I^B	I^BI^B	B
I^B	i	I^Bi	B
i	i	ii	O

Blood Group Shows → Law of Dominance, Multiple allels, Law of Segrigation, Co-dominance.

Second Law ↷ Law Of Segregation

By Monohybrid →

*During gamete formation, two allels of gene must separate to move into different gametes.

*Also Called Law of purity of gametes. ★

*No exception to this law.

Tt

T t → pure gametes

* INHERITANCE OF 2 GENES / 2 CHARACTERS / DIHYBRID :→

Y → Dominant Yellow seed Colour
y → Recessive green seed colour
R → Round Shaped Seeds
r → wrinkled shaped seeds

RRYY X rryy

Round Yellow Seeds | Wrinkled green seeds

RY | ry gametes

F1 gen. RrYy Dihybrid, all round Yellow

Selfing → RrYy X RrYy

	RY	Ry	rY	ry
RY	RRYY	RRYy	RrYY	RrYy
Ry	RRYy	RRyy	RrYy	Rryy
rY	RrYY	RrYy	rrYY	rrYy
ry	RrYy	Rryy	rrYy	rryy

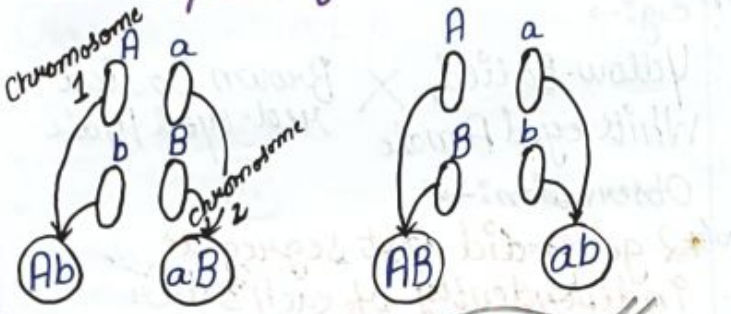
phenotypic Ratio : 9:3:3:1 ★★

Genotypic Ratio : 1:2:1:2:4:2:1:2:1 ★

Third Law → Law of Independent Assortment.

↳ Based on Dihybrid cross.

"When two pairs of traits are combined in hybrid, segregation of one pair of characters is independent of other pair of characters."



Exception to this law → Linkage

*Chromosomal Theory of Inheritance :→

Mendel published his Work = 1865
Year of Rediscovery = 1900

* Why his work remain hidden for years?

1. Communication was not easy.
2. His work could not be widely publicised.
3. Concept of factors, (pair of alleles don't show blending) was not accepted.
4. Use of mathematics and statistics was unaccepted.
5. Very advanced as of that time.

In 1900 de Vries, Correns and Von Tschermak independently rediscovered Mendel's result.

In 1902, Chromosome movement during meiosis has been worked out.

Advancement in Microscopy
↓
Carefully observed cell division
↓
led to discovery of chromosomes
(advanced bodies visualised by staining)
↓
These double & divide before cell division

Behaviour of Chromosomes	Behaviour of Genes
Occur in pairs	Occur in pairs
Segregate at the time of gamete formation such that only one of each pair is transmitted to a gamete.	Segregate at gamete formation and only one of each pair is transmitted to a gamete.
Independent pair segregate independently of each other.	One pair segregates independently of another pair.

Meiosis I → Anaphase

two chromosome pairs can align at metaphase plate independent of each other.

Walter Sutton and Theodore Boveri

1. They noted that behavior of chromosome was ∥ to behavior of genes and used chromosome movement to explain Mendel's Law.
2. They argued → pairing and separation of pair of chromosomes would lead to segregation of pair of factors they carried.
3. Sutton United Knowledge of Chromosomal segregation with Mendel's principle and called it Chromosomal Theory of Inheritance. ★

Thomas Hunt Morgan

1. Father of experimental Genetics.
2. He discovered basis of Variation produced in Sexual reproduction. ★
3. He worked with Tiny fruit flies. Drosophila Melanogaster. ★

Tiny Fruit Flies

1. Could be grown on Simple Synthetic medium in Laboratory.
2. Complete their Life cycle in 2 weeks.
3. Single mating produce large no. of progeny.
4. Clear differentiation in male and female sexes.
5. Has many types of Heredity Variation that can be seen with low power microscope.

Linkage and Recombination

Morgan → dihybrid crosses in Drosophila (coined term Linkage) ↓ to study genes that were Sex-linked.

e.g.:→ Yellow-Bodied, White eyed Female × Brown bodied red-eyed Male

Observation:→

- 2 genes did not segregate independently of each other
- Genes were located on X-chromosome.

Dihybrid Cross

Two genes on same Chromosome ↓ proportion of parental gene combination is more than non parental type. ★

Why? → Due to Linkage. ↓ linkage refers to physical association of genes on chromosom ★

Recombination

Generation of Non-parental gene combinations. ★

- Recombination can be high or low

e.g.→ (1) ★ White and Yellow Tightly Linked 1.3% recombination

e.g.→ (2) White & Miniature Wing loosely linked 37.2% recombination

Recombination never > 50%

Alfred Sturtevant

Used frequency of recombination b/w gene pairs on same chromosome as a measure of distance b/w genes and mapped their position on the chromosome. → 'Genetic Maps'.

Cross A → 0.1%

♀ X ♂
Yellow white — Wild type

↓

Linkage - 98.7%
Recombination - 1.3%

Cross B →

♀ X ♂
white miniature — Wild type

↓

Linkage - 62.8%
Recombination - 37.2%

NEET SLAYER

Polygenic Inheritance

When more than 1 gene pair control a single character.

e.g. → Human Height, Human Skin Color.

- Traits are expressed across a gradient.
- also takes into account, influence of environment.
- effect of each allel is additive.

e.g. → AABBCC → AaBBCC → AaBbCC → AaBbCc → aaBbCc → aabbCc → aabbcc

AABBCC (darkest skin colour)
aabbcc (lightest skin colour)

Pleiotropy

- Single Gene → Multiple Phenotypic expression.
  → pleiotropic Gene.
- Gene affects various metabolic pathways, and contributes towards different phenotypes.

Example-1 :→ phenylKetonuria.

phenylalanine —(phynl alanine hydrolase)→ Tymosine.

① Gene for phenylalanine hydrolase get muted.
② Mental Retardation, Reduction in Hair and Skin Pigment.

Example-2 :→

Character 1	BB	Bb	bb
Shape of Seed	○	○	◌
Character 2			
Size of starch grain	Large	Intermediate	Small

Sex Determination

Cytological observations made in a no. of insects led to development of concept of genetic/chromosomal basis of sex-determination.

Henking (1891) ⭐ → trace a specific nuclear structure all through spermatogenesis in a few insects.

- He observed that 50% sperm received this structure after spermatogenesis, whereas 50% sperm did not receive it.
- He named this structure → X body.
- X-body - Chromosome (by further investigation) X-chromosome.

X - Chromosomes → Sex Chromosome
Rest Chromosomes → Autosomes

Male Heterogamety

XX-XY
- Humans
- Drosophila
- Some Insects

XX → Female
XY → Male

XX-XO
- Males have 1 less chromosome than female.
- e.g. → Grasshopper ⭐

XX → Female
XO → Male

Female Heterogamety

ZZ-ZW
- e.g. → Birds

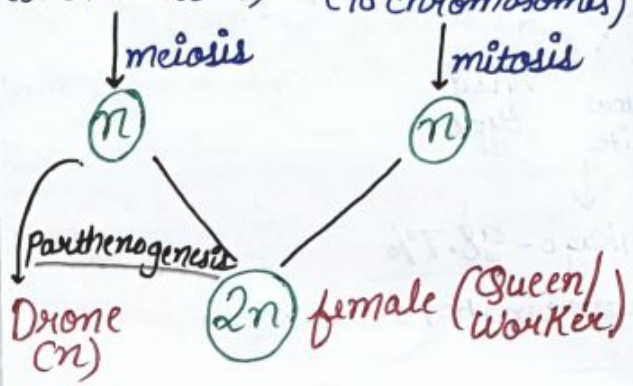
ZW → Female

Sex Determination In Honey Bee

→ Based on no. of sets of chromosomes an individual receives.

→ Haplodiploid ⭐ sex determination system.

Female (2n) (32 chromosomes) X Drone (n) (16 chromosomes)



Mutation

results in:
- Alternation of DNA sequences
- Changes in genotype and phenotype of organism

Mutation, Recombination = Variation

① Lass (deletation)
② Gain (Insertion/Duplication)
of segment of DNA result in alternation of chromosome.
↓
abnormalities/aberrations.
↓
Commonly in Cancer cells

① Point Mutation → Change in single base pair of DNA. ⭐
e.g. → Sickle Cell Anemia.
② Frame Shift Mutation → Deletions/Insertions of Base pairs of DNA.

Mutagens → Chemical & physical factors that induces mutations.
e.g. → UV radiations.

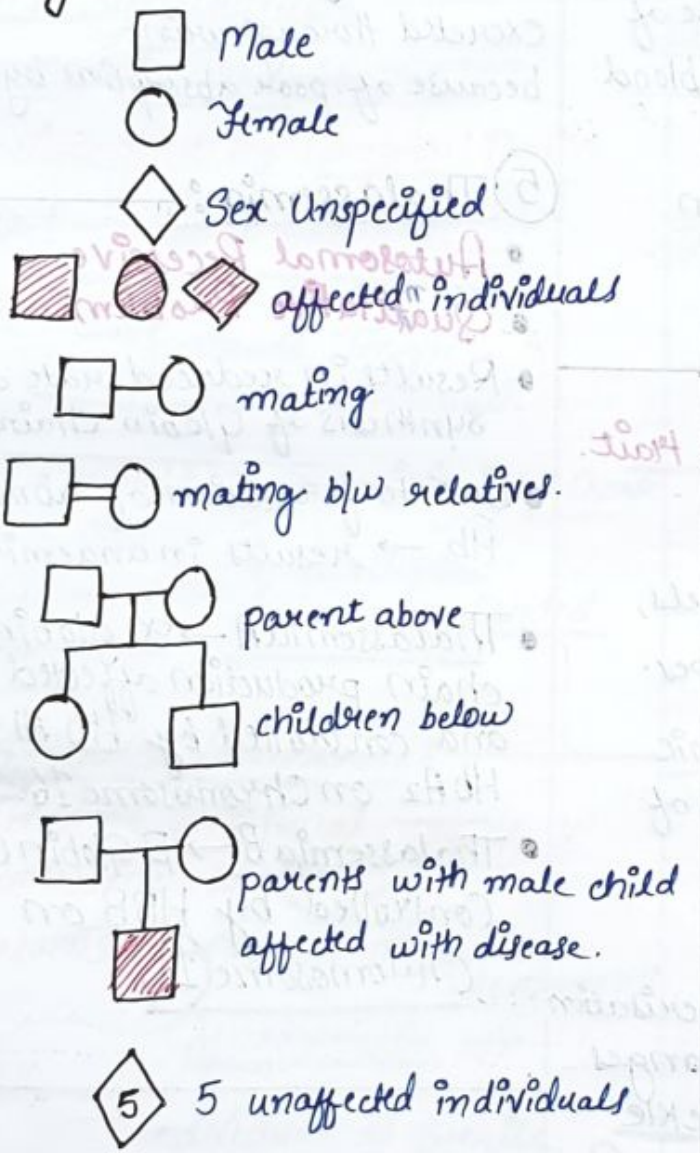
* Genetic Disorders :→

* Pedigree Analysis :

Study of family history about inheritance of particular trait provides alternative. Such analysis of traits in several generations of family is pedigree analysis.

- Pedigree is used to trace →
- inheritance of specific trait, abnormality of disease.

:→ Symbols used in Human pedigree analysis :→



Clues to Solve Pedigree :→

(1) X-linked recessive →
→ Criss Cross Inheritance
→ Affects male (majority)
eg → Colour Blindness, Haemophilia, Duchene muscular dystrophy

(2) Autosomal Dominant →
→ effect equal in male and female in every generation.
e.g. → Myotonic Dystrophy, Huntington's Disease.

(3) Autosomal Recessive →
→ Homozygous Condition.
→ Either both parents should be carrier, or one parent affected and other carrier.
e.g. → PKU, Thallesemia, Sickel Cell Anaemia.

* Mendelian Disorders :

- Due to alternation / mutation in single gene.
- Transmitted to offsprings on Mendel's inheritance basis.
- Can be traced by pedigree analysis.
- Can be dominant, recessive, or sex linked.

(1) Colour Blindness :→

- Sex linked recessive Disorder.
- Due to genes present on X-chromosome
- Due to defect in red / green cone.
- Occur in 8% male and 0.4% female.

Case 1 → Mother (Carrier) X Father (Colour Blind)

Son and Daughter Can be Colour Blind (50%-50%)

Case 2 → Mother (Colour Blind) X Father (Normal)

Son (Color Blind) and Daughter (50% Chances)

(2) Haemophilia:

- Sex-Linked recessive Disorder
- Results in non stop Bleeding
- A single protein, (part of cascade of protein) involved in clotting of blood is affected.
- eg → Family pedigree of Queen Victoria.

(3) Sickle Cell Anaemia:

- Autosomal Linked recessive trait.
- Qualitative Problem.
- Controlled by single pair of alleles, Hb^A and Hb^S → possible genotypes.
- Due to substitution of glutamic acid by Valine at 6th position of β-Globin chain. (Imp)
- GAG → GUG. (Imp)
- Mutant Hb undergoes polymerisation under low O_2 tension and changes RBC's from Biconcave to Sickle like structure.

(4) Phenyl Ketonuria:

- Autosomal recessive
- Inborn error of Metabolism
- Affected individual lacks phenylalinine hydroxylase
- Phenylalanine
  - result → gets accumulated
  - converts → Tyrosine
    - Phenylpyruvic acid & other derivatives → excreted through urine because of poor absorption by Kidn
    - Leads to mental retardatio

(5) Thalassemia:

- Autosomal Recessive
- Quantitative Problem.
- Results in reduced rate of synthesis of Globin chains (α & β)
- Due to formation of abnormal Hb → results in anaemia.
- Thalassemia A → α globin chain production affected and controlled by Hb A1 and Hb A2 on Chromosome 16. (Imp)
- Thalassemia B → B Globin Chain. Controlled by HBB on Chromosome 11.

* Chromosomal Disorders :

Due to absence/excess/abnormal arrangement of chromosome.

* Aneuploidy :→ Failure of segregation of chromosome.

* Polyploidy :→ Failure of cytokinesis after telophase which results in an increase in whole set of chromosome.
  - Often seen in plants.

① Down's Syndrome :

- Trisomy of chromosome 21.
- First discovered by Langdon Down.

Result : → Short Statured.
→ Small Round Head.
→ Furrowed Tongue.
→ Partially open mouth.

→ Palm Broadt Characteristic Palm Crease

→ Physical
→ Psychomortor } developed → retarded.
→ mental

② Klinefelter's Syndrome :

- Presence of additional copy of 'X' chromosome i.e. XXY.

Symptoms : → Overall Masculine development
→ Gynaecomastia also.
→ Individual is Sterile.

③ Turner's Syndrome :

- Due to absence of one X chromosome i.e. 45 with XO.

→ Female → Sterile, (Rudimentary ovaries)
→ Lack of other secondary sexual characters.