5. PRINCIPLES OF INHERITANCE AND VARIATION

MENDEL'S LAWS OF INHERITANCE

Gregor Mendel is the Father of genetics.
He conducted some hybridization experiments on **garden peas (*Pisum sativum*).**

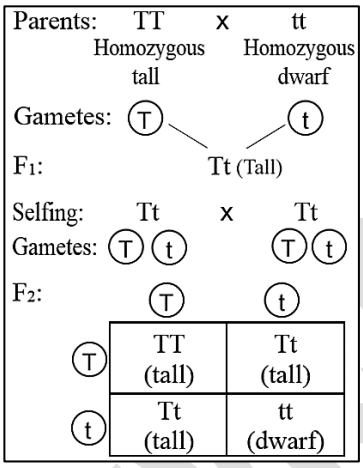
Mendel selected 7 pairs of true breeding pea varieties:

7 Characters	Contrasting Traits	
	Dominant	Recessive
1. Stem height	Tall	Dwarf
2. Flower colour	Violet	White
3. Flower position	Axial	Terminal
4. Pod shape	Inflated	Constricted
5. Pod colour	Green	Yellow
6. Seed shape	Round	Wrinkled
7. Seed colour	Yellow	Green

INHERITANCE OF ONE GENE

Monohybrid cross: A cross involving 2 plants differing in one character pair. E.g. Mendel crossed tall and dwarf pea plants to study the inheritance of one gene.

- **Allele:** Alternative forms of a gene. E.g. T (tall) and t (dwarf) are two alleles of a gene for the character height.
- **Phenotype:** Physical expression of a character.
- **Genotype:** Genetic constitution of a character.



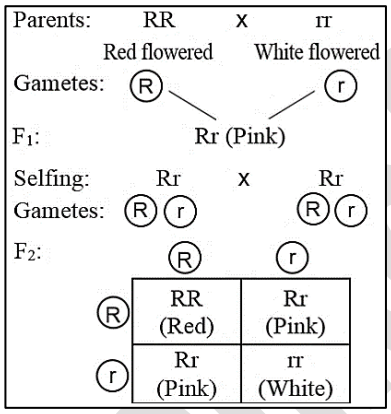
Monohybrid phenotypic ratio:
3 Tall: 1 Dwarf = **3:1**

Monohybrid genotypic ratio:
1 Homozygous tall (TT)
2 Heterozygous tall (Tt)
1 Homozygous dwarf (tt)
= **1:2:1**

OTHER PATTERNS OF INHERITANCE

1. Incomplete Dominance

- It is an inheritance in which heterozygous offspring shows intermediate character b/w two parental characteristics.
- E.g. Flower colour in **snapdragon (dog flower or *Antirrhinum sp.*)** and ***Mirabilis jalapa* (4'O clock plant).**



Here, cross between homozygous **red** & **white** produces **pink** flowered plant. Thus phenotypic & genotypic ratios are same.

Phenotypic ratio=
1 Red: 2 Pink: 1 White

Genotypic ratio=
1 (RR): 2 (Rr): 1(rr)

2. Co-dominance

- It is the inheritance in which both alleles of a gene are expressed in a hybrid. E.g. ABO blood grouping in human.
- ABO blood groups are controlled by the gene **I**.
- This gene controls the production of **sugar polymers (antigens)** that protrude from plasma membrane of RBC.
- The gene **I** has three alleles I^A, I^B & **i**.
- I^A and I^B produce a slightly different form of the sugar while allele **i** doesn't produce any sugar.

Alleles from parent 1	Alleles from parent 2	Genotype of offspring	Blood types (phenotype)
I^A	I^A	$I^A I^A$	A
I^A	I^B	$I^A I^B$	AB
I^A	i	I^Ai	A
I^B	I^A	$I^A I^B$	AB
I^B	I^B	$I^B I^B$	B
I^B	i	I^Bi	B
i	i	ii	O

When I^A and I^B are present together, they both express their own types of sugars. This is due to **co-dominance.**

CHROMOSOMAL THEORY OF INHERITANCE

Proposed by **Walter Sutton** & **Theodore Boveri.**

Thomas Hunt Morgan proved chromosomal theory of inheritance using fruit flies ***(Drosophila melanogaster)***.

It is the suitable material for genetic study because,

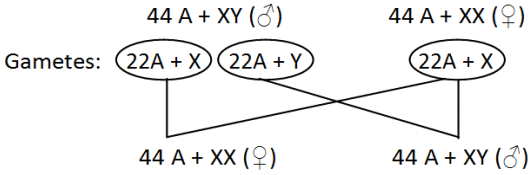
- They can grow on simple synthetic medium.
- Short generation time (life cycle: 12-14 days).
- Breeding can be done throughout the year.
- Hundreds of progenies per mating.
- Male and female flies are easily distinguishable. E.g. Male is smaller than female.
- It has many types of hereditary variations that can be seen with low power microscopes.

SEX DETERMINATION

Sex Determination in Humans (XX-XY type)

- Human has **23 pairs** of chromosomes (22 pairs of autosomes and 1 pair of sex chromosomes).
- A pair of X-chromosomes **(XX)** is present in the **female,** whereas **X and Y** chromosomes are present in **male.**
- During spermatogenesis, males produce 2 types of gametes: 50 % with X-chromosome and 50 % with Y-chromosome.

- Females produce only ovum with an X-chromosome.
- There is an equal probability of fertilization of the ovum with the sperm carrying either X or Y chromosome.
- The sperm determines whether the offspring male or female.



PEDIGREE ANALYSIS

- In human, control crosses are not possible. So the study of family history about inheritance is used.
- Such an analysis of genetic traits in several generations of a family is called **pedigree analysis**.
- The representation or chart showing family history is called **family tree (pedigree).**
- In human genetics, pedigree study is utilized to trace the inheritance of a specific trait, abnormality or disease.

Symbols used in pedigree analysis

Male: □ Female: ○ Sex unspecified: ◇

Affected individual: ■ ● ◆ Mating: □—○

Mating b/w relatives (consanguineous mating): □═○

Parents above & children below

Parents with affected male child

Five unaffected offspring

GENETIC DISORDERS

1. Mendelian Disorders

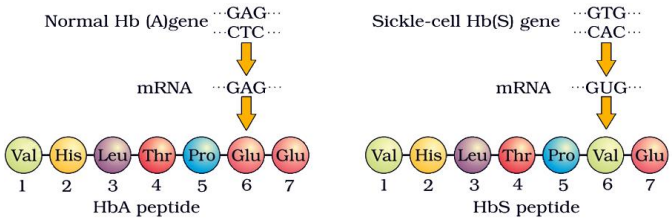
Haemophilia (Royal disease):

- It is a sex linked (X-linked) recessive disease.
- In this, a protein involved in the blood clotting is affected.
- A simple cut results in non-stop bleeding.
- The disease is controlled by 2 alleles, **H** & **h**. **H** is normal allele and **h** is responsible for haemophilia.
- In females, haemophilia is very rare because it happens only when mother is at least carrier and father haemophilic.
- Queen Victoria was a carrier of hemophilia. So her family pedigree shows many haemophilic descendants.

Sickle-cell anaemia:

- This is an autosome linked recessive disease.
- It can be transmitted from parents to the offspring when both the partners are carrier (heterozygous) for the gene.
- The disease is controlled by a pair of allele, Hb^A and Hb^S.

  Homozygous dominant (Hb^AHb^A): normal
  Heterozygous (Hb^AHb^S): carrier; sickle cell trait
  Homozygous recessive (Hb^SHb^S): affected
- The defect is caused by the substitution of **Glutamic acid (Glu)** by **Valine (Val)** at the **sixth position** of the **β-globin chain** of the haemoglobin (Hb).
- This is due to the single base substitution at the sixth codon of the **β-globin gene** from **GAG** to **GUG.**
- The mutant Hb molecule undergoes polymerization under low oxygen tension causing the change in shape of the RBC from biconcave disc to elongated sickle like structure.



2. Chromosomal disorders

- **Down's syndrome:** It is the presence of an additional copy of chromosome number 21 **(trisomy of 21).**

  Genetic constitution: 45 A + XX or 45 A + XY (i.e. **47** chromosomes).

  Features:
  - They are short statured with small round head.
  - Broad flat face.
  - Furrowed big tongue and partially open mouth.
  - Many "loops" on finger tips.
  - Broad palm with characteristic palm simian crease.
  - Retarded physical, psychomotor & mental development.
  - Congenital heart disease.
- **Klinefelter's Syndrome**: It is the presence of an additional copy of X-chromosome in male (trisomy).

  Genetic constitution: 44 A + XXY (i.e. **47** chromosomes).

  Features:
  - Overall masculine development. However, the feminine development is also expressed. E.g. Development of breast **(Gynaecomastia).**
  - Sterile.
  - Mentally retarded.
- **Turner's syndrome**: This is the absence of one X chromosome in female (monosomy).

  Genetic constitution: 44 A + X0 (i.e. **45** chromosomes).

  Features:
  - Sterile, Ovaries are rudimentary.
  - Lack of other secondary sexual characters.
  - Dwarf.
  - Mentally retarded.

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