PRINCIPLES OF INHERITANCE AND VARIATION

Introduction:

- **Genetics** is the study of genes, genetic code, inheritance and variation of characters from parents to offspring.
- **Inheritance** is the process by which characters or traits are transferred from parents to progeny.
- **Variation** is the degree by which progeny differs from each other and with their parents.

Mendel's Experiment:

- **Gregor Johann Mendel** is called as "Father of genetics". He performed hybridization experiments by taking 7 contrasting characters of 14 true-breeding **garden pea** (*Pisum sativum*) plants and proposed the laws of inheritance.
- He selected garden pea plant because it has many distinct contrasting characteristics, easily available and grown on a large scale, can be self and cross-pollinated, etc.
- Mendel called genes as **factors**, which are passed from parents to offsprings and the genes that code for a pair of opposite traits is called **alleles.**

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Character	Dominant trail	Recessive trait
Stem length	 Tall	 Dwarf
Pod shape	 Inflated	 Constricted
Seed shape	 Round	 Wrinkled
Seed colour	 Yellow	 Green
Flower position	 Axial	 Terminal
Flower colour	 Purple	 White
Pod colour	 Green	 Yellow

Inheritance of One Gene (Monohybrid Cross):

➔ In monohybrid cross, two organisms differing in one single trait at a time is crossed. **Example:** a cross between tall and dwarf pea plants, considering the trait of plant height only.

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MONOHYBRID CROSS

Tall Plant Dwarf Plant } Parents

TT X tt

}Gametes

(T) (t)

}F_1 (Heterozygous Tall)

Tt

Tt X Tt }F_1 on Sel n g

(T) (t) (T) (t) }Gametes

Checker Board

♂ / ♀	(T)	(t)
(T)	TT Homozygous Tall	Tt Heterozygous Tall
(t)	Tt Heterozygous Tall	tt Homozygous Dwarf

Monohybrid Cross Ratio

Phenotypic ratio : 3: 1 (3 Tall: 1 Dwarf)

Genotypic ratio: 1: 2:1 (1TT: 2 Tt: 1 tt)

- **Law of dominance** (first law) states that in a heterozygous condition, the allele whose characters are expressed over the other is called **dominant** (appears in F_1 generation) and the characters that remain hidden are called **recessive** (appears in F_2 generation).
- **Law of Segregation** (second law) states that during gamete formation, the factors or alleles of a character

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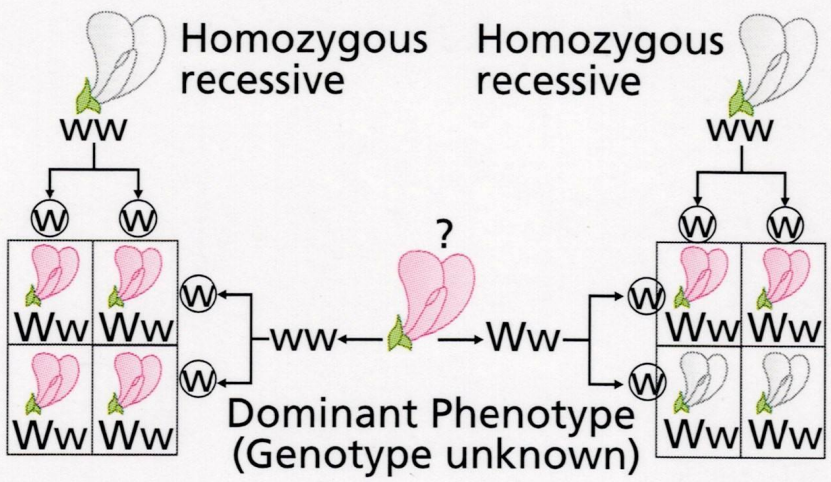
pair present in parents segregate from each other such that a gamete receives only one of the two factors.

Back Cross and Test Cross:

- **Back cross** is the cross between a hybrid of F_1 progeny with one of their parents.
- **Test cross** is the cross between a tall plant (dominant) from F_2 generation with a dwarf plant (homozygous recessive parent). Mendel performed this cross to find out that the dominant genotype is homozygous or heterozygous. **Example:**



All owers are violet

Unknown ower is homozyous dominant

Half of the flowers are violet and half of the flowers are white

Unknown ower is heterozygous

Inheritance of Two Gene (Dihybrid Cross):

- In dihybrid cross, two organisms differing in two traits at a time is crossed. **Example:** a cross between a pea

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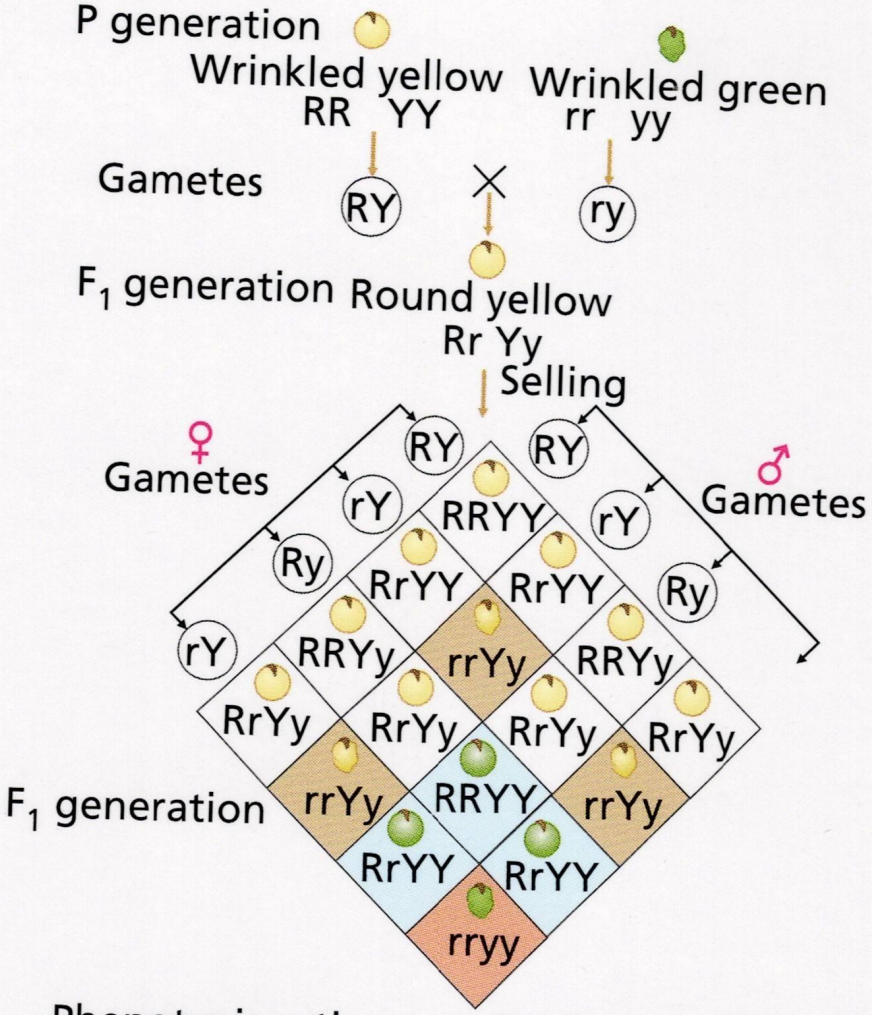
plant with yellow colour and round shaped seeds and the other having green colour and wrinkled seeds.



Phenotypic ratio: round yellow: round green:
9 3
wrinkled yellow: wrinkled green
3 1

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- Law of independent assortment states that 'when two pairs of traits are combined in a hybrid, segregation of one pair of characters is independent of the other pair of characters'.

Chromosomal Theory of Inheritance:

- In 1900, **de Vries, Carl Correns** and **von Tschermak** worked independently and rediscovered Mendel's results.
- In 1902, **Theodore Boveri** and **Walter Sutton** independently gave this theory. They said that the pairing and separation of a pair of chromosomes would lead to segregation of a pair of factors (gene) they carried. According to this theory:
  - Genes are located on chromosomes which are vehicles of heredity.
  - Two identical chromosomes form a homologous pair. They segregate during meiosis in the process of gamete formation.
  - Chromosomes segregate as well as assort independently.
- **Thomas Hunt Morgan** extensively worked on fruit flies, *Drosophila melanogaster* and provided experimental evidence to support the chromosomal theory of inheritance.

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Linkage and Recombination:

- Linkage is the physical association of two or more genes on a chromosome. In a dihybrid cross, if the two genes are tightly linked or present on the same chromosome, the parental combination is more prevalent than non-parental combinations (recombination).
- The linkage and recombination are directly dependent on the distance between a pair of genes. More the distance, greater is the probability of recombination.
- Morgan carried out several dihybrid crosses in *Drosophila* to study sex-linked genes. Two of them are given on next page.
- **Alfred Sturtevant** used the frequency of recombination between gene pairs on the same chromosome to measure the distance between genes and also 'mapped' their position on the chromosome.

$$\text{Distance between genes} = \frac{\text{Total Number of Recombinants}}{\text{Total Number of Offsprings}}$$

Sex Determination:

- In 1891, **Henking** first observed X chromosome and named it as X body.
- In 1905, EB Wilson and Stevens named XY chromosomes as **Allosomes** or **Sex chromosomes**. The rest of the chromosomes are termed as **Autosomes.**

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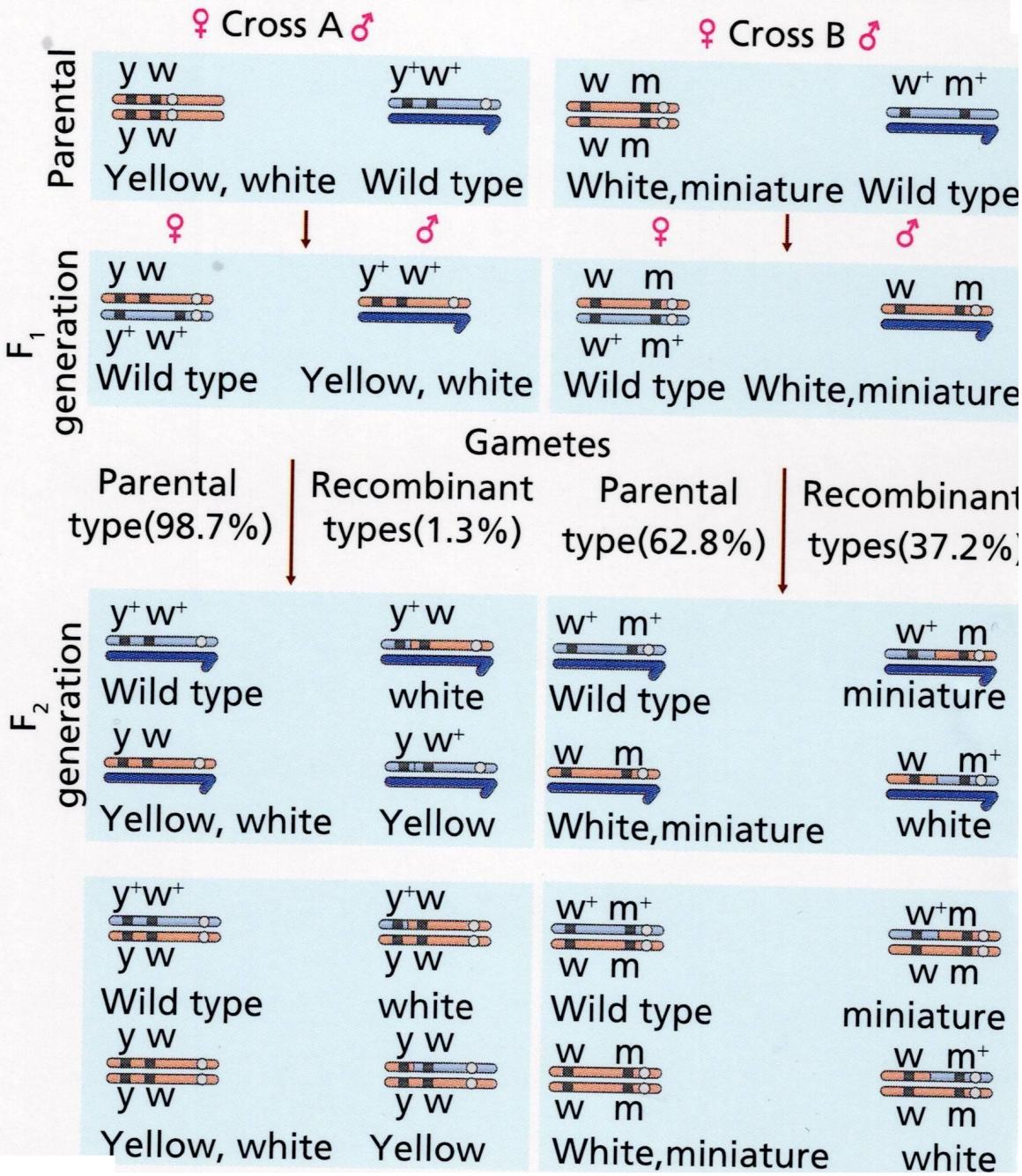

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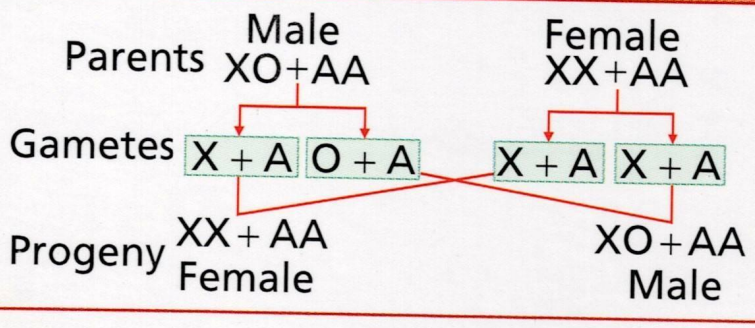
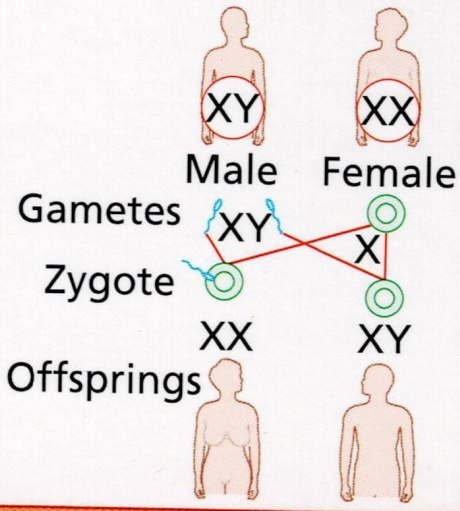
Male heterogamety

XX–XO mechanism
e.g., Grass-hoppers

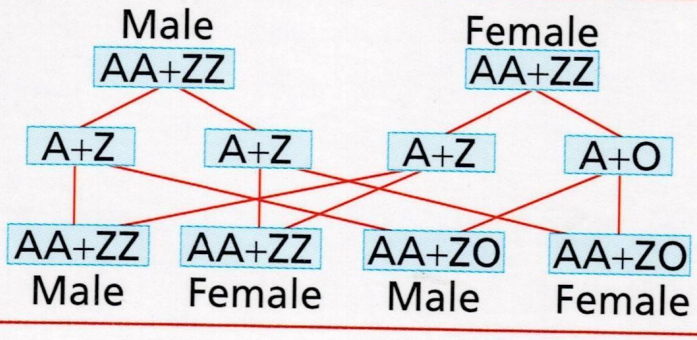


XX–XY mechanism
e.g., Humans, *Drosophila*



Female heterogamety

ZZ–ZO mechanism
e.g., Butterflies



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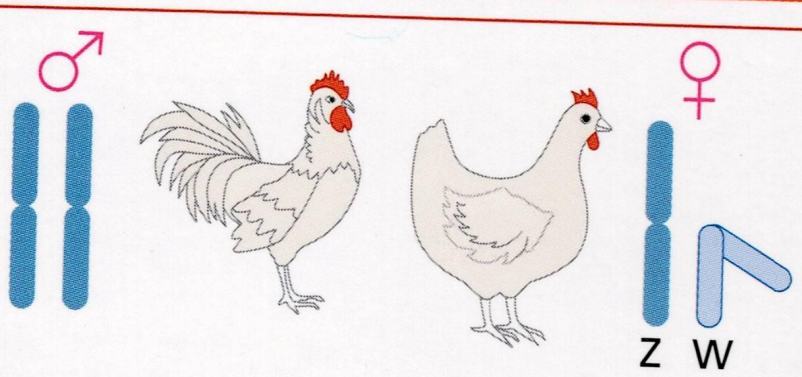
ZZ–ZW mechanism
e.g., Birds



Male bird
AA+ZZ

X

Female bird
AA+ZW

A+Z (sperm) A+Z (sperm)

A+W (ova) A+Z (ova)

A+Z
A+W

A+Z	AA+ZZ (Male)	AA+ZW (Female)
A+Z	AA+ZZ (Male)	AA+ZW (Female)

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Haplodiploid method	
Haploid-diploid mechanism e.g., Honeybees	Female (Queen) 2n = 32 (Diploid) → Meiosis → Eggs → (n = 16) → Parthenogenesis → Haploid male (n = 16) Male (Drone) n = 16 (Haploid) → Mitosis → Sperm → (n = 16) → Fertilization → Diploid female (n = 32) They do not have Father but have Grandfather (Maternal).

Mutation:

➔ It is a sudden heritable change in DNA sequences that resulting in changes in the genotype and phenotype of an organism. It is induced by mutagens such as UV radiation, formaline, etc.

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- There are two types of **gene mutations:**
  - **Point mutation:** Mutation due to change in a single base pair of DNA e.g., sickle cell anemia.
  - **Frame-shift mutation:** Mutation due to loss (deletions) or gain (insertion/duplication) of a base pair of DNA.
- In **chromosomal aberrations**, there is loss or gain of a segment of DNA that results in structural alterations in chromosomes and also led to different abnormalities.
- **Genomic** or numerical mutations are of two types:
  - **Aneuploidy:** Loss or gain of one or more chromosomes due to failure of segregation of homologous pair of chromosomes or chromatids during meiosis (cell division). It is sub-divided into monosomy and trisomy.
  - **Polyploidy:** A whole set of chromosomes in an organism is increased as a result of failure of cytokinesis after telophase stage of cell division. It is sub-divided into autopolyploidy and allopolyploidy.

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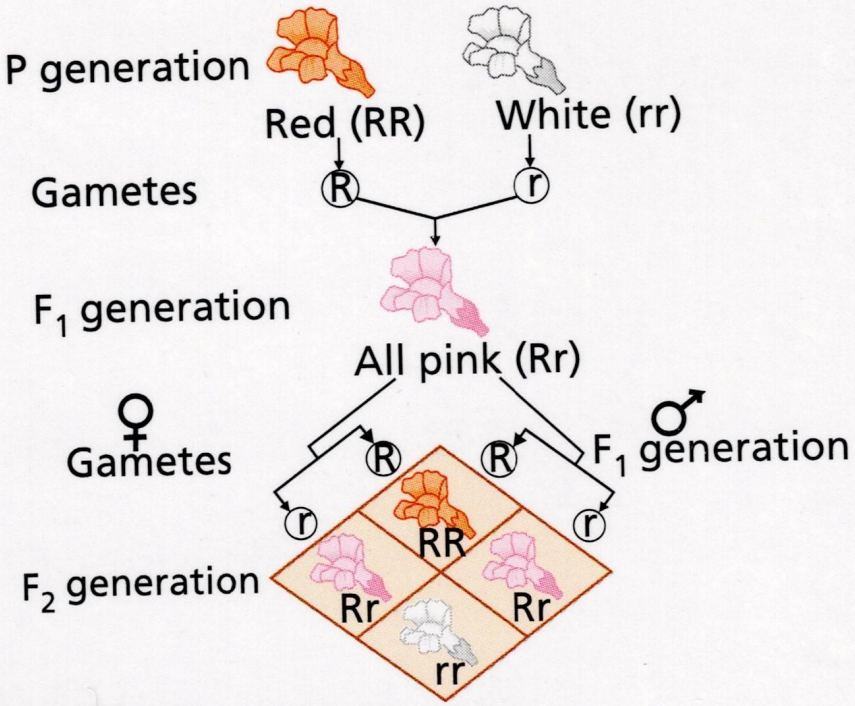
Deviations from Mendel's Laws:

Incomplete dominance: (Blending Inheritance)

- It is a phenomenon in which the phenotype of the heterozygous offsprings does not resemble either of the two parents. Instead, it shows an intermediate character. **Example:** Inheritance of flower colour in Snapdragon or *Antirrhinum majus* (dog flower) and in *Mirabilis jalapa.*



Phenotypic ratio: red : pink : white
1 : 2 : 1

Genotypic ratio: RR: Rr: rr
1 : 2 : 1

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Co-dominance:

- It is a phenomenon in which two alleles express themselves independently when present together in an organism. **Example:** ABO blood grouping in humans.
- ABO blood groups are controlled by the **gene I** which has three alleles I^A, I^B and i/I^O. Out of these, I^A and I^B are completely dominant over i.

Table Showing the Genetic Basis of Blood Grouping in Human Population

Allele from Parent 1	Allele from Parent 2	Genotype of Offspring	Blood-types of Offspring
I^A	I^A	$I^A I^A$	A
I^A	I^B	$I^A I^B$	AB
I^A	*i*	I^A i	A
I^B	I^A	$I^A I^B$	AB
I^B	I^B	$I^B I^B$	B
I^B	*i*	I^B i	B
i	*i*	*ii*	O

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Multiple allelism:

- It is the presence of more than two alleles of a gene (on the same locus on a chromosome) that governs the same character. **Example:** ABO blood grouping in humans.

Pleiotropy:

- When a single gene controls multiple phenotypic traits, is known as a pleiotropic gene. The different phenotypes are mostly a result of the effect of a gene on metabolic pathways. **Example:** starch synthesis in pea, phenylketonuria, etc.

Genotype	Phenotypes	
	Size of starch grains	Seed shape
BB	Large sized	Round
Bb	Intermediate size	Round
bb	Small	Wrinkled

- In phenylketonuria, single gene mutation produces many phenotypic effects such as mental retardation, reduction in hair and skin pigmentation.

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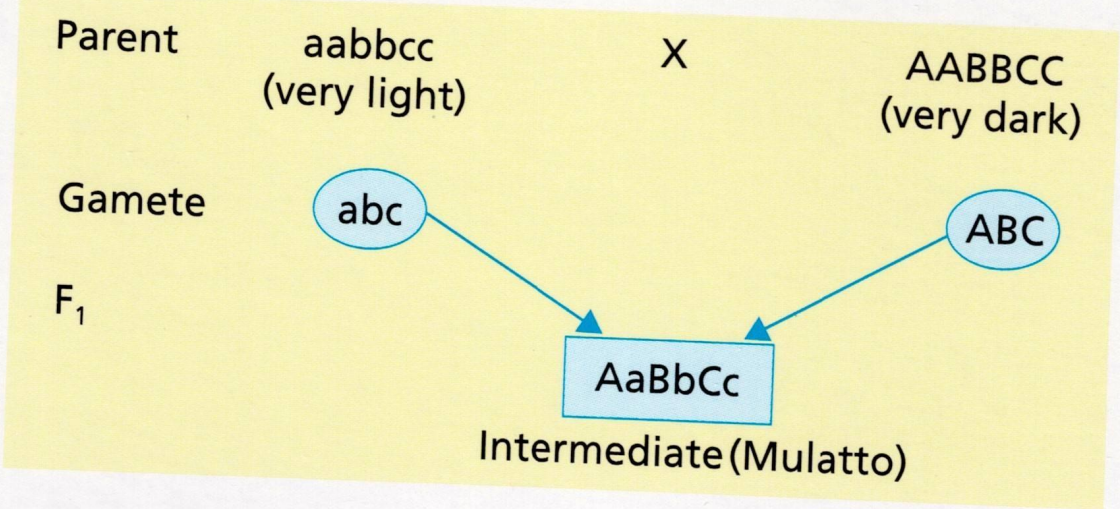
Polygenic inheritance:

- Here, a trait is governed by multiple independent genes. It is also affected by environmental conditions. **Example:** human skin colour, height, etc.



Pedigree Analysis:

- It is analysis of inheritance of a particular trait through several generations in a family. The representation or chart showing family history is called **family tree** (pedigree).
- It provides a strong tool to trace the inheritance of a specific trait, abnormality or disease.

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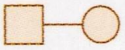
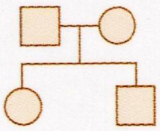
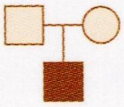
- Symbols used in the pedigree analysis are:

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 

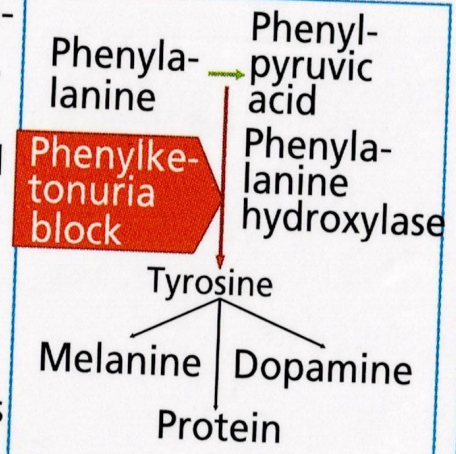
Genetic Disorders:

- These can be grouped into two types, i.e., Mendelian disorders and Chromosomal disorders.
- **Mendelian disorders** are usually caused by alteration or mutation in the single gene. These disorders can be dominant or recessive.

Mendelian disorders	Genetic Trait	Cause	Effects	Inheritance pattern and Features
Haemophilia	X-linked recessive	Defect in protein involved in the clotting of blood	Continuous bleeding from wounds	The heterozygous carrier female (X^hX) may transmit the disease to sons. The possibility of a female becoming a haemophilic (X^hX^h) is very rare, for this mother has to be at least carrier and father should be haemophilic.
Colour blindness	X-linked recessive	Defect in the green or red cone of the eye	Unable to discriminate between red and green colour	A daughter will be colour blind only if the mother is a carrier and father is colour blind. There is a 50 percent probability of a carrier female to transfer the disease to sons.

Sickle cell anaemia	Auto-some -linked recessive	Single base substitution of Glutamic acid (Glu) by Valine (Val) at the 6^{th} position of the beta-globin chain of haemoglobin.	The mutant Hb undergoes polymerization under low O_2 tension causing the change in shape of the RBC to sickle structure. Thus, RBCs destroyed rapidly and leads to anaemia.	Offsprings may get the disease when both the parents are a carrier or heterozygous (Hb^AHb^S).

Thalas-semia	Auto-some-linked recessive	Production of α globin chain is affected (α **Thalassemia)** and mutation occurs in the HBA1 and HBA2 genes present on the chromo-some 16 or production of β globin chain is affected (β **Thalassemia)** and mutation occurs in the HBB gene present on the chromosome 11.	Formation of abnor-mal hae-moglobin molecule resulting in anae-mia	Offsprings may get the disease when both the parents are a carrier (heterozy-gous).

Phenylketonuria	Autosome-linked recessive	Lack of phenylalanine hydroxylase enzyme that converts phenylalanine to tyrosine	Mental retardation. Accumulation and excretion of phenylalanine and its derivatives in urine	
Cystic fibrosis	Autosome-linked recessive	Mutation occurs on the 7th chromosome	Body produces abnormal glycoproteins that interfere with the salt metabolism.	The mucus secreted by body becomes abnormally sticky and viscid and therefore blocks the passage in the lungs, liver and pancreas.

- **Chromosomal disorders** are usually caused by absence or excess or abnormal arrangement of one or more chromosomes. These can be easily studied by analysis of Karyotypes.

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Chromo-somal disorders	Karyotype/ Genetic constitution	Symptoms
Down's syndrome (Mongol-oidiocy)	45 A + XX / 45 A + XY (trisomy of chromosome 21)	• Mental retardation, Heart defects. • Short stature, small round head and broad flat face. • Partially open mouth with furrowed tongue. • Broad palm with palm crease.
Klinefelter's syndrome	44 A + XXY (trisomy of X-chromosome in male)	• Sterile male • Overall masculine development with some feminine characters (development of breast, i.e., **Gynaecomastia**). • Feminine pitched voice and poor beard growth.
Turner's syndrome	44 A + XO (Monosomy of X–chromosome in female)	• Sterile (Poorly developed ovaries and sex glands) • Lack of secondary sexual characters. • Short stature, small uterus, puffy fingers and webbed neck.

TEST

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1. Identify and write the correct statement:
   (a) *Drosophila* male has one X and one Y chromosome.
   (b) *Drosophila* male has two X chromosomes.
   (c) *Drosophila* male has one Z and one W chromosome.
   (d) *Drosophila* male has two Z chromosomes.
2. The autosomal disorder/disease in humans is:
   (a) Colour blindness
   (b) Thalassemia
   (c) Haemophilia
   (d) Turner's Syndrome
3. In his classic experiment on peas, Mendel did not use:
   (a) Seed shape
   (b) Seed colour
   (c) Pod Length
   (d) all of the above
4. The F_2 generation of a cross produced identical phenotypic and genotypic ratio. It is not an expected Mendelian result, and can be attributed to:
   (a) Independent assortment
   (b) Linkage
   (c) Incomplete dominance
   (d) All of the above

TEST

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Solutions:

1. **Option (a) is correct.**

   XX-XY type of mechanism of sex-determination is found in *Drosophila melanogaster.* In this, males have autosomes along with one X and one Y chromosome. Whereas, females in this have autosomes along with pair of XX chromosomes.

2. **Option (b) is correct.**

   Thalassemia is an autosome-linked recessive blood disease in humans. This disease transmitted from parents to the offspring both the parents are carrier (heterozygous) for the gene.

3. **Option (c) is correct.**

   In classic experiment on peas, Mendel use seven contrasting characters such as stem length, pod shape, seed shape, seed color, flower position, flower color and pod color.

4. **Option (c) is correct.**

   In incomplete dominance, the F_2 generation of a cross produced identical phenotypic and genotypic ratio, i.e., 1:2:1. It is considered as the deviation from mendelian results, in which the dominant allele does not completely express itself. Thus, heterozygous offsprings produce an intermediate character.

TEST

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5. In Mendelian dihybrid cross, when heterozygous round yellow are self-crossed, round green offsprings are produced which are represented by genotype?

   (a) RrYy, RrYY, RRYy
   (b) RrYY, RRyy, rryy
   (c) rrYy, rrYY
   (d) RRyy, Rryy

Practice Time

6. A mutation is a:
   (a) sudden non-inheritable change in an organism's genetic material.
   (b) change in phenotype followed by a change in genotype.
   (c) change in hereditary material directed by a changing environment.
   (d) change in genotype which may result in a new expression of a characteristic.

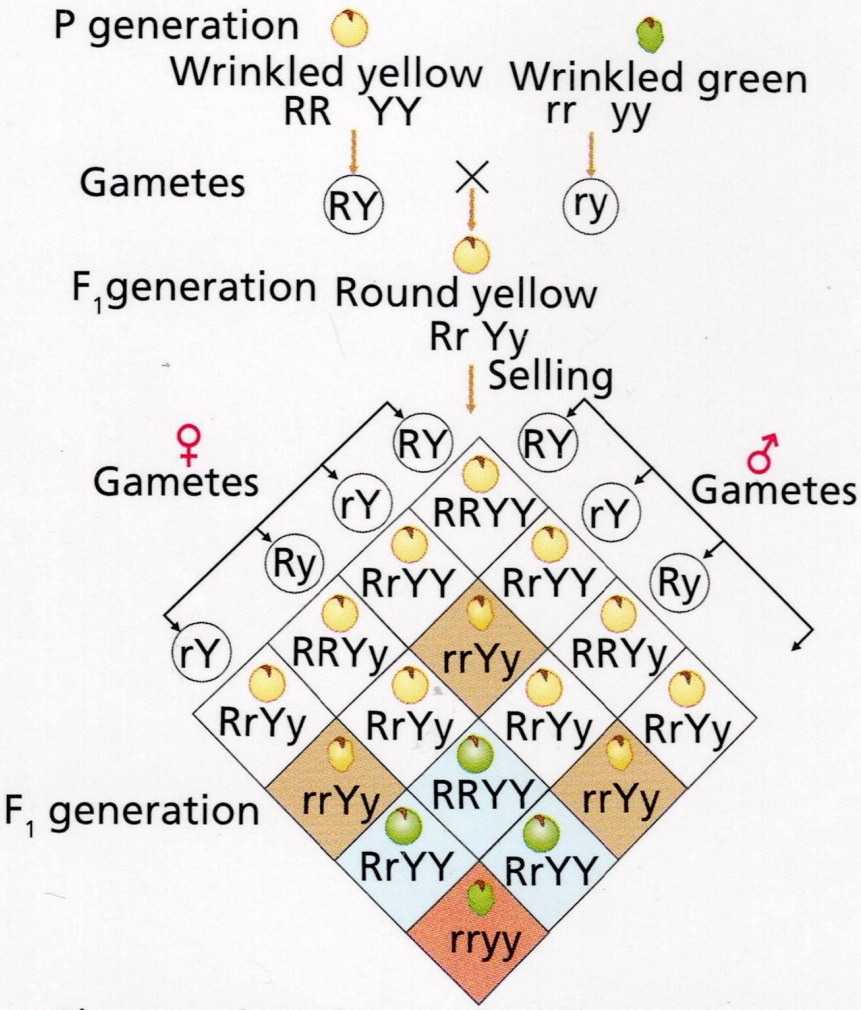
Option (d) is correct.

7. The genotypes of a husband and wife are I^AI^B and I^Ai. Among the blood types of their children, how many different genotypes and phenotypes are possible?
   (a) 4 genotypes; 4 phenotypes
   (b) 3 genotypes; 3 phenotypes
   (c) 3 genotypes; 4 phenotypes
   (d) 4 genotypes; 3 phenotypes

Option (d) is correct.

Solutions:

5. Option (d) is correct.



Phenotypic ratio: round yellow: round green: wrinkled yellow: wrinkled green

9 : 3 : 3 : 1