ZLGY-MM: XII

5. PRINCIPLES OF INHERITANCE & VARIATION

- ♥ **Heredity:** Resemblance b/w offspring and their parents.
- ♥ **Inheritance:** Transmission of characters from parents to progeny.
- ♥ **Variation:** Difference between parents and offspring.
- ♥ **Genetics:** Study of heredity and variation. (William Bateson-1905)

Mendel's Laws of Inheritance

- ➢ **Gregor Mendel** set-up a basic framework of rules governing inheritance, therefore known as **Father of genetics**.
- ➢ He conducted hybridisation experiments on garden peas –*Pisum sativum*- for 7 years (1856-63)

Mendel selected 7 pairs of true breeding pea varieties:-

Characters	Dominant	Recessive
1. Stem height	Tall (110 cm)	Dwarf (30 cm)
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

- ➢ Mendel's experiment were a great success because:-
  a) Statistical analysis and mathematical logic were applied.
  b) Large sampling size, gave credibility to the data that he collected.
- ♥ Reasons for selection of *Pisum sativum*:-
  i. Plant shows clear cut contrasting characters
  ii. Easy to cultivate
  iii. Fertile hybrids
  iv. Floral structure is suitable for artificial pollination
  v. Short growth period & life cycle
  vi. A large number of offspring are produced.

TERMS USED IN MENDELIAN GENETICS

- ♥ **Character:** It is a feature of the individual. E.g. Stem height
- ♥ **Trait:** An inherited character. E.g. Tall or dwarf
- ♥ **Gene:** The **segment of DNA** that determines a particular trait. Mendel called it as ***factors***.
- ♥ **Alleles:** The alternative forms of a gene. E.g. T (tall) and t (dwarf) are two alleles of a gene responsible for the character height.
- ♥ **Homozygous:** The condition in which chromosome carries similar alleles for a character. E.g. TT, yy etc.
- ♥ **Heterozygous:** The condition in which chromosome carries dissimilar alleles for a character. E.g. Tt, Yy etc.
- ♥ **Dominant character:** The character which is expressed in heterozygous condition or in F_1 generation. It indicates with capital letter.
- ♥ **Recessive character:** The character which is suppressed in heterozygous condition or in F_1 generation. It indicates with small letter.
- ♥ **Phenotype:** Physical (visible) expression of an individual. E.g. Tall, dwarf etc.
- ♥ **Genotype:** Genetic make-up of an individual. E.g. TT, tt etc.
- ♥ **Wild type:** The species variety showing normal phenotype.
- ♥ **Hybrid:** An individual produced by the mating of genetically unlike parents.
- ♥ **Punnett square/Checker board:** A grid that enables to calculate the results of genetic crosses. (By R.C. Punnett, British geneticist).
- ♥ **Cross:** Deliberate mating of 2 parental types.

  Steps in making a cross in pea:
  I. Selection of 2 pea plants with contrasting characters.
  II. Consider one as female parent & remove anthers (*emasculation*) of plant to avoid self pollination.
  III. Collection of pollen grains from the other plant (male parent) and transferred to female parent (pollination).
  IV. Collection of seeds and production of offspring.
- ♥ **Backcross** and **Testcross**
  - **Backcross:** Crossing of F_1 hybrid with its any of parent.
  - **Testcross:** Crossing of an F_1 hybrid with its recessive parent. It is used to find out the unknown genotype.

  N.B.:- Confusion exists only in case of dominant phenotype, whether it is due to homozygous dominant or heterozygous genotype.
  - Recessive phenotype only expresses when genotype is homozygous recessive.

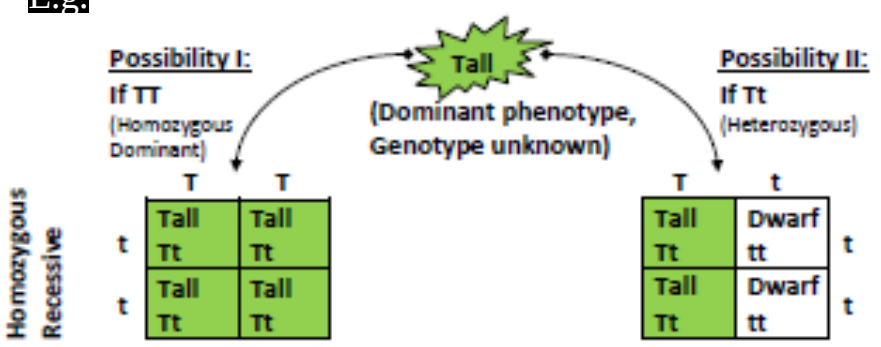
Procedure for Test cross -

The individual with unknown genotype is crossed with its recessive parent.

Case 1st. ➡ If all the progeny are dominant, then the unknown genotype is homozygous dominant.

Case 2nd. ➡ If half of the progeny are dominant and half recessive (1:1), then the unknown genotype is heterozygous.

E.g.



INHERITANCE OF ONE GENE (Monohybrid Cross)

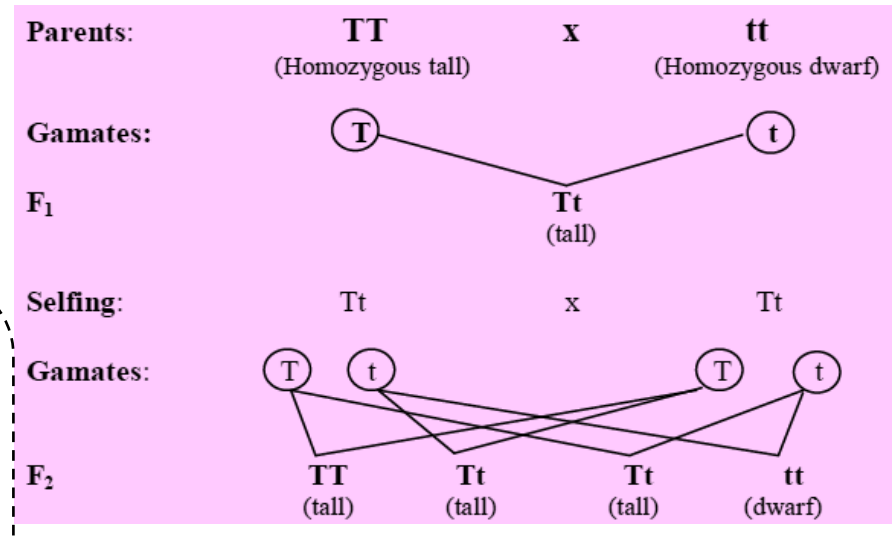
Monohybrid cross: A cross involving 2 plants differing in one character pair.

E.g. Mendel crossed tall and dwarf pea plants to study the inheritance of one gene.

Result:-

Phenotypic ratio: 3 Tall: 1 Dwarf= **3:1**

Genotypic ratio: 1 TT: 2 Tt:1 tt= **1:2:1**



Tt x Tt

The F_1 (Tt) when self pollinated, produces gametes **T** and **t** in equal proportion. During random fertilization, pollen grains of **T** have **50%** chance to pollinate eggs of **T** & **t**. Also, pollen grains of **t** have **50%** chance to pollinate eggs of **T** and **t.**

- $1/4^{th}$ of the fertilization leads to TT (¼ TT).
- $2/4^{th}$ (1/2) of the fertilization leads to Tt (½ Tt).
- $1/4^{th}$ of the fertilization leads to tt (¼ tt).

This ratio is mathematically condensable to form the binomial expression $(ax + by)^2$

Hence $(½\ T + ½\ t)^2$ = (½ T + ½ t) (½ T + ½ t)

= ¼ TT + ½ Tt + ¼ tt

Mendel's Principles/ **Laws of Inheritance:-**

1st -*Law of Dominance*

"When a pair of contrasting characters combines, only one is expressed (dominant character) and the other remains hidden (recessive character)".

3 connotations of this law
- *Characters are controlled by discrete units called **factors.***
- *Factors occur in pairs.*
- *In a dissimilar pair of factors one member of the pair dominates **(dominant)** the other **(recessive).***

The concept of dominance

In heterozygotes, there are dominant and recessive alleles.

The normal (unmodified or functioning) allele of a gene produces a normal enzyme that is needed for the transformation of a substrate.

The modified allele is responsible for production of

(i) The normal/less efficient enzyme or
(ii) A non-functional enzyme or
(iii) No enzyme at all

In the first case: The modified allele will produce the same phenotype like unmodified allele. It becomes dominant.

In 2nd and 3rd cases: The phenotype will dependent only on the functioning of the unmodified allele. Here, the modified allele becomes recessive.

2nd -*Law of Segregation*

"During gamete formation, the factors (alleles) of a character pair present in parents do not mix each other, but separate and segregate from each other such that a gamete receives only one of the 2 factors".

- Homozygous parent produces similar gametes.
- Heterozygous parent produces two kinds of gametes each having one allele with equal proportion.

INHERITANCE OF TWO GENES (Dihybrid cross)

Dihybrid cross: A cross between two parents differing in 2 pairs of contrasting characters.

E.g. Cross b/w pea plant with round shaped & yellow coloured seeds (RRYY) and wrinkled shaped & green coloured seeds (rryy).

➔ On observing the F_2, Mendel found that the yellow and green colour segregated in a 3:1 ratio. Round and wrinkled seed shape also segregated in a 3:1 ratio.

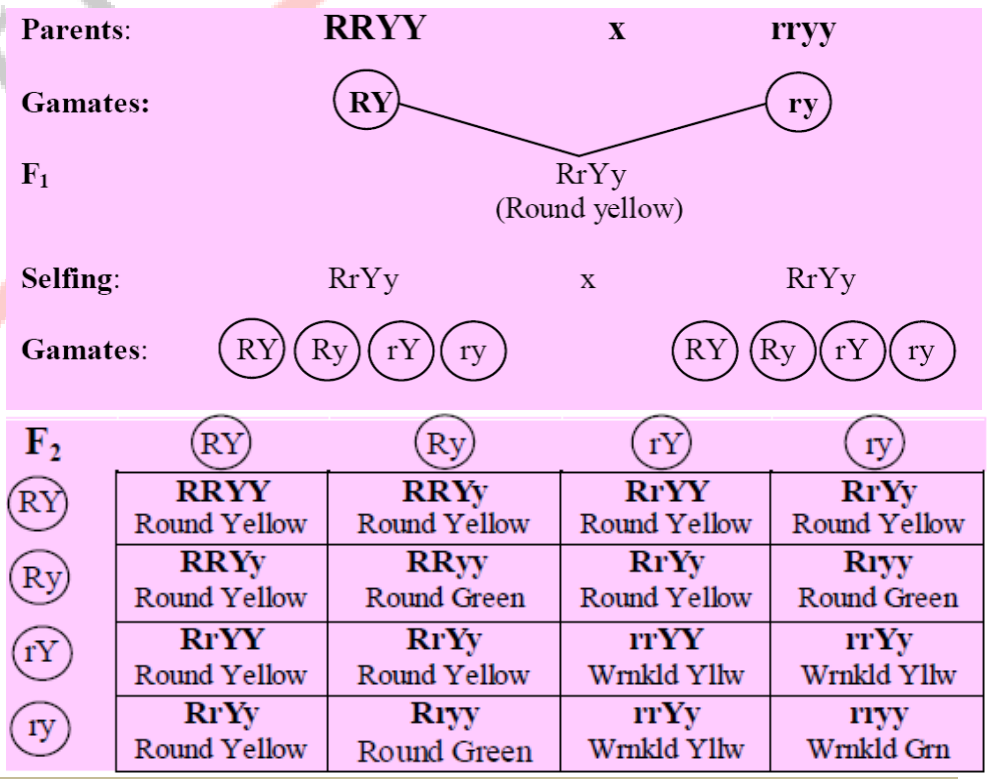
Result:-

Phenotypic ratio= 9 Round yellow: 3 Round green: 3 Wrinkled yellow: 1 Wrinkled green= **9:3:3:1**

The ratio of 9:3:3:1 can be derived as a combination series of 3 yellow: 1 green, with 3 round: 1 wrinkled. i.e. **(3: 1) (3: 1) = 9: 3: 3: 1**

Dihybrid genotypic ratio: RRYY =1 RRYy =2 RRyy =1 RrYY =2 RrYy =4 Rryy =2 rrYY =1 rrYy =2 rryy =1

i.e., **1:2:1:2:4:2:1:2:1**

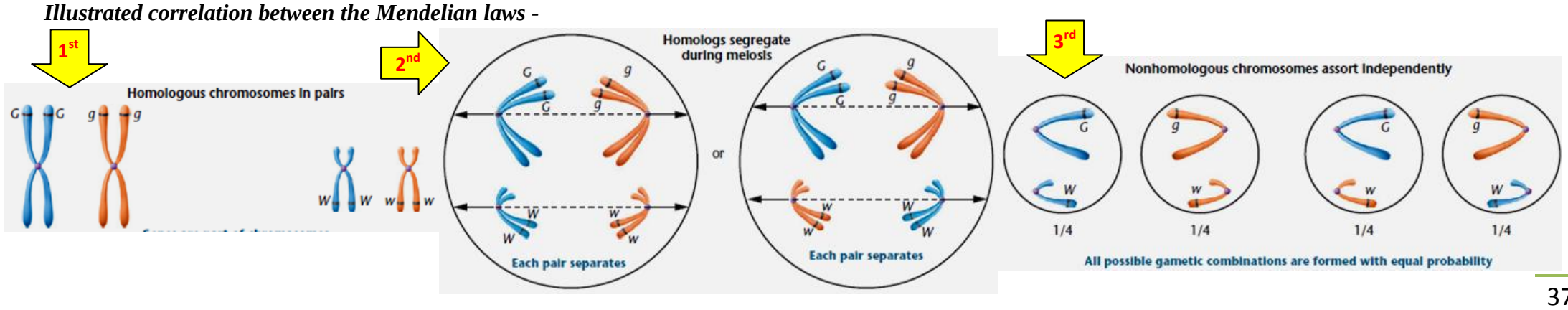


F_2	RY	Ry	rY	ry
RY	**RRYY** Round Yellow	**RRYy** Round Yellow	**RrYY** Round Yellow	**RrYy** Round Yellow
Ry	**RRYy** Round Yellow	**RRyy** Round Green	**RrYy** Round Yellow	**Rryy** Round Green
rY	**RrYY** Round Yellow	**RrYy** Round Yellow	**rrYY** Wrnkld Yllw	**rrYy** Wrnkld Yllw
ry	**RrYy** Round Yellow	**Rryy** Round Green	**rrYy** Wrnkld Yllw	**rryy** Wrnkld Grn

3rd - *Law of Independent Assortment*:

"When more than one pair of characters are involved in a cross, factor pairs independently segregate from the other pair of characters'.

Illustrated correlation between the Mendelian laws -



NON-MENDELIAN INHERITANCE

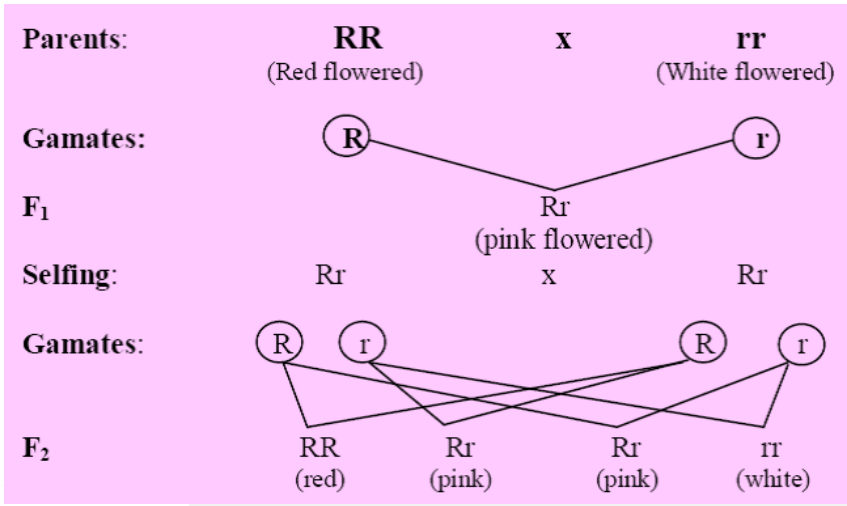
➔ *These are inheritance which do not obey Mendelian laws*

1. Incomplete Dominance

- *It is an inheritance in which heterozygous offspring shows intermediate character b/w two parental characteristics.*

E.g. 1. Flower colour in **snapdragon (dog flower or *Antirrhinum sp.*)**
2. Flower colour in ***Mirabilis jalapa* (4'O clock plant).**

- Here the gene for red colour is incompletely dominant over the white and formation of a new colour (pink) is the interaction between these two.



Result:-

Phenotypic ratio= 1 Red: 2 Pink: 1 White

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

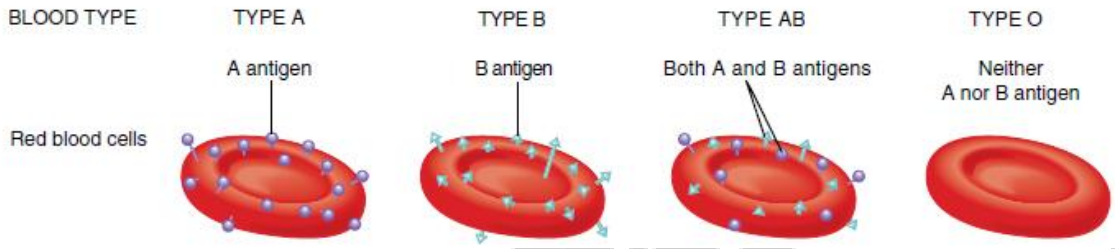
2. Co-dominance

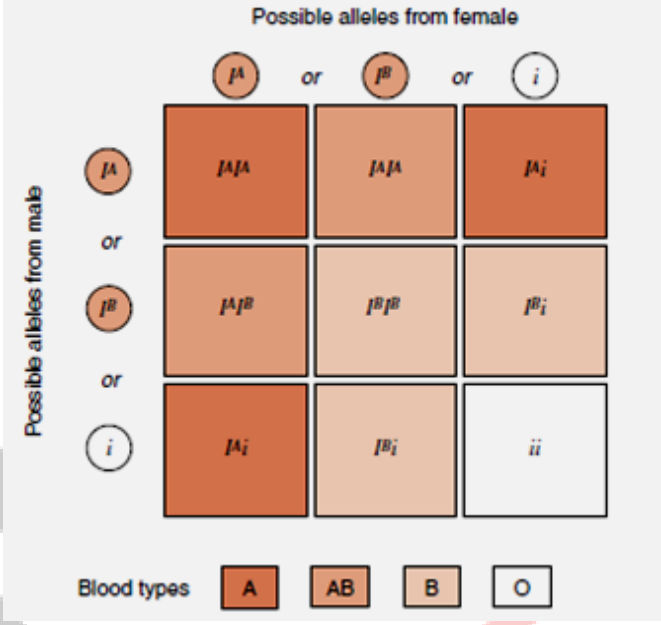
- *It is the inheritance in which both alleles of a gene are expressed in a hybrid.*

E.g. ABO blood grouping in human.

- ABO blood groups are controlled by the gene ***I***. The plasma membrane of the RBC has sugar polymers that protrude from its surface and is controlled by the gene.
- The gene ***(I)*** has three alleles I^A, I^B and ***i***. The alleles I^A and I^B produce a slightly different form of the sugar while allele ***i*** doesn't produce any sugar.

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





3. Multiple allelism

- *It is the inheritance in which more than 2 alleles govern the same character.*

E.g. ABO blood grouping (3 alleles: I^A, I^B & *i*).

4. Pleiotropy

- *It is the inheritance in which a single gene governs multiple phenotypic expressions. Such a gene is called* ***pleiotropic gene****.*

E.g. Starch synthesis in pea seeds, sickle cell anaemia, phenylketonuria etc.

Starch synthesis in pea plant:

- Starch is synthesized effectively by **BB** and therefore, large starch grains are produced.
  bb have lesser efficiency in starch synthesis and produce smaller starch grains.
  But, **Bb** produce an intermediate-sized starch grains.
- If **starch grain size** is considered as phenotype, the alleles show **incomplete dominance.**

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

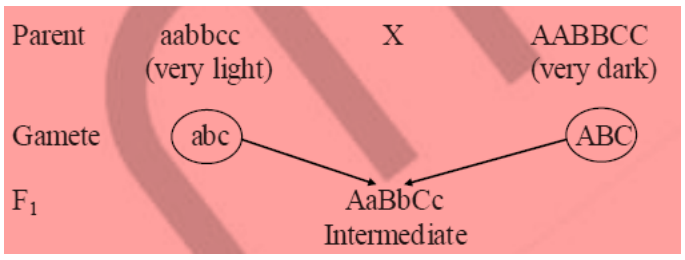
5. Polygenic Inheritance

- *It is the inheritance in which some traits are controlled by 3 or more genes (multiple genes).*
  - Polygenic inheritance also takes into account the influence of environment.
  - In a polygenic trait, the phenotype reflects the contribution of each allele, i.e., the *effect of each allele is additive*.

E.g. Human skin colour, human height, flower colour etc.

Human Skin Colour

- Assume that 3 genes A, B, C control skin colour in human.
- The dominant forms **A**, **B** and **C** responsible for *dark* skin colour and the recessive forms **a**, **b** and **c** for *light* skin colour.
  - Genotype with all the dominant alleles (AABBCC) = darkest skin colour
  - Genotype with all the recessive alleles (aabbcc) = lightest skin colour.
  - Genotype with 3 dominant alleles + 3 recessive alleles (AaBbCc)= intermediate colour.



F_2

Gametes	ABC	ABc	AbC	aBC	Abc	aBc	abC	abc
ABC	**AABBCC** Very dark	**AABBCc** Dark	**AABbCC** Dark	**AaBBCC** Dark	**AABbCc** Fairly Dark	**AaBBCc** Fairly Dark	**AaBbCC** Fairly Dark	**AaBbCc** Intermediate
ABc	**AABBCc** Dark	**AABBcc** Fairly Dark	**AABbCc** Fairly Dark	**AaBBCc** Fairly Dark	**AABbcc** Intermediate	**AaBBcc** Intermediate	**AaBbCc** Intermediate	**AaBbcc** Fairly light
AbC	**AABbCC** Dark	**AABbCc** Fairly Dark	**AAbbCC** Fairly Dark	**AaBbCC** Fairly Dark	**AAbbCc** Intermediate	**AaBbCc** Intermediate	**AabbCC** Intermediate	**AabbCc** Fairly light
aBC	**AaBBCC** Dark	**AaBBCc** Fairly Dark	**AaBbCC** Fairly Dark	**aaBBCC** Fairly Dark	**AaBbCc** Intermediate	**aaBBCc** Intermediate	**aaBbCC** Intermediate	**aaBbCc** Fairly light
Abc	**AABbCc** Fairly Dark	**AABbcc** Intermediate	**AAbbCc** Intermediate	**AaBbCc** Intermediate	**AAbbcc** Fairly Light	**AaBbcc** Fairly Light	**AabbCc** Fairly light	**Aabbcc** Light
aBc	**AaBBCc** Dark	**AaBBcc** Intermediate	**AaBbCc** Intermediate	**aaBBCc** Intermediate	**AaBbcc** Fairly Light	**aaBBcc** Fairly Light	**aaBbCc** Fairly light	**aaBbcc** Light
abC	**AaBbCC** Dark	**AaBbCc** Intermediate	**AabbCC** Intermediate	**aaBbCC** Intermediate	**AabbCc** Intermediate	**aaBbCc** Intermediate	**aabbCC** Fairly Light	**aabbCc** Light
abc	**AaBbCc** Intermediate	**AaBbcc** Intermediate	**AabbCc** Fairly light	**aaBbCc** Fairly light	**Aabbcc** Light	**aaBbcc** Light	**aabbCc** Light	**aabbcc** White

CHROMOSOMAL THEORY OF INHERITANCE

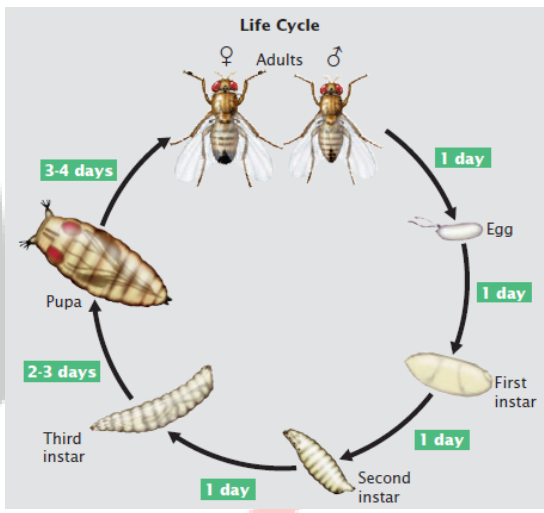
- Mendel's work was described in a paper "*Experiment on plant hybridisation"* (1865). But it remained unrecognized till 1900 because,
  1. Communication was not easy.
  2. His concept of **genes** (**factors)** as **stable** and **discrete** units was not accepted, because continuous variation seen in nature.
  3. His **mathematical approach** was new and **unacceptable**.
  4. Mendel could **not provide any physical proof** for the existence of factors.
- In 1900, **de Vries** (Holland)**, Correns** (Germany) & **von Tschermak** (Austria) independently rediscovered Mendel's results.
- **Chromosomal Theory (1902): Theodore Boveri** studied the *chromosomal movement* during meiosis. He noted that the behaviour of chromosome were parallel to the behaviour of genes. **Walter Sutton** united chromosomal segregation with Mendelian principles and called it the **chromosomal theory of inheritance**.

A comparison between the behaviour of chromosome and the gene.

CHROMOSOME	GENE
1. Occurs in pairs 2. They segregate at the time of gamete formation such that only one of each pair is transmitted to a gamete. 3. Independent pairs segregate independently of each other.	1. Occurs in pairs 2. They segregate at the time of gamete formation such that only one of each pair is transmitted to a gamete. 3. One pairs segregate independently of another pair.

PRINCIPLES-

- ✓ Genes are located on chromosomes which are vehicles of heredity. They are transmitted from parents to offspring, i.e. they are immortal.
- ✓ Two identical chromosomes form a homologous pair. They segregate at the time of gamete formation.
- ✓ Independent pairs segregate independently of each other.
- ✓ Chromosomes are mutable.



T.H Morgan proved chromosomal theory of inheritance using fruit flies (***Drosophila melanogaster)***. It is the suitable material because,

a. It breeds very quickly and breeding could be done through-out the year.
b. Short generation time (life cycle: 12-14 days)
c. Hundreds of progenies per mating. Each female lay 400-500 eggs
d. They can grow on simple synthetic medium.
e. Male and female flies are easily distinguishable.
f. It has many types of hereditary variations that can be seen with low power microscopes.
g. 4 pair of large chromosomes (avoid complications).

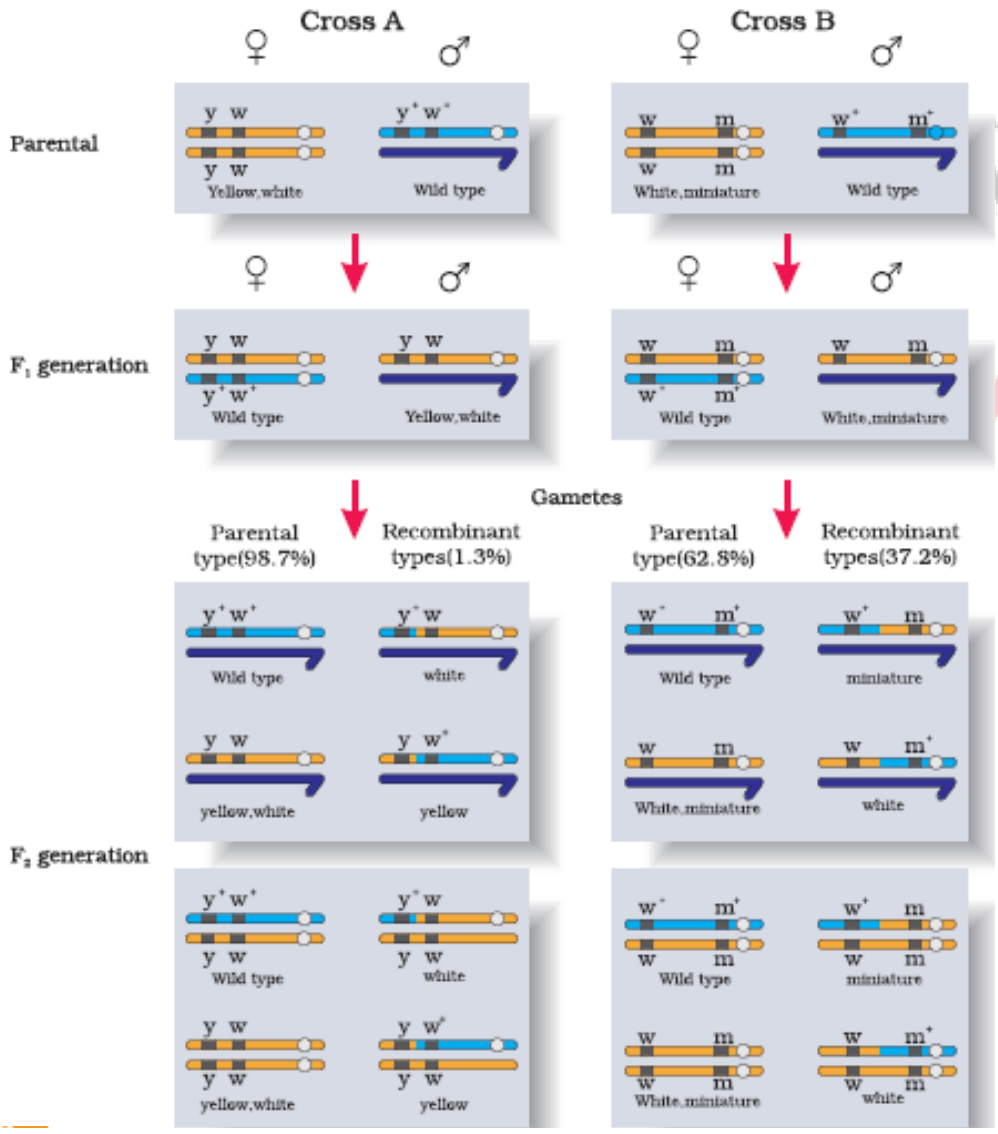
Linkage and Recombination

- ➢ **Linkage:** Physical association of 2 or more genes on a chromosome. They do not show independent assortment.
- ➢ **Recombination:** It is the generation of non-parental gene combinations.

Morgan carried out several dihybrid crosses in ***Drosophila*** to study sex-linked genes.

Cross A
Yellow-bodied(y), white-eyed (w) female
X
Brown-bodied(y^+), red-eyed male (w^+)-wild type

Cross B
White-eyed(w), miniature winged (m)female
X
Red eyed(w^+), large winged male(m^+)-wild type



Morgan's Conclusion-

- ✓ When two genes were situated on the same chromosome, the proportion of parental gene combinations was much higher than the non-parental type. This is due **linkage.**
- ✓ **Tightly linked genes** show **low recombination**. **Loosely linked** genes show **high recombination.**

GENETIC MAP

- **Genetic map:** *Linear graphic representation of the sequence and relative distances of the genes present in the chromosome.* It is based on the recombination frequency.
- **Alfred Sturtevant** (student of Morgan-1911) constructed the first genetic map.

Sex Determination

➔ Chromosome is the main factor determining the sex of an organism.

There are 2 types of chromosomes in an organism-

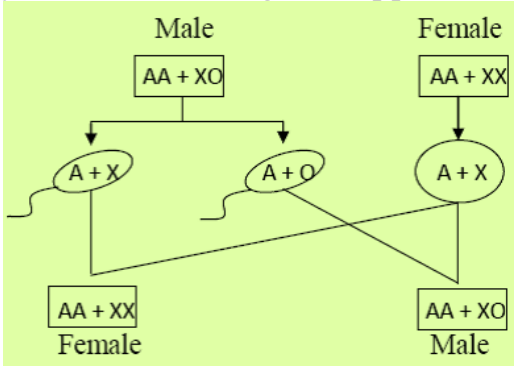
1. **Autosomes** are chromosomes which determine the somatic characters of the organism.
2. **Sex chromosomes** are the chromosomes which involve in sex determination.

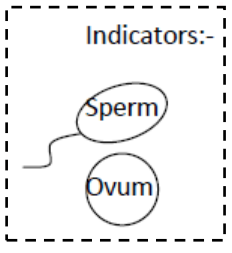
- Studies carried out by **Henking (1891)** revealed the existence of sex chromosome. He studied spermatogenesis in some insects and observed that 50 % of sperm received a nuclear structure after spermatogenesis, whereas other 50 % sperm did not receive it. Henking called this structure as the **X body** (later it is called as **X-chromosome)**.

Mechanism of sex determination

a. XX female –XO male mechanism: Here, male is heterogametic, i.e. XO (produce gametes with X and gametes without X) and female is homogametic, i.e. XX (all gametes are with X chromosomes).

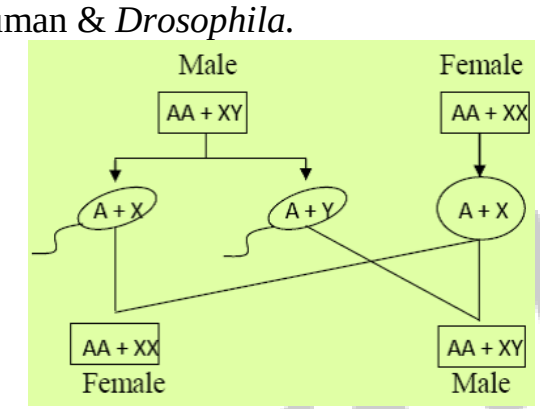
E.g. Many insects such as grasshopper, cockroach, spider, bedbug etc.

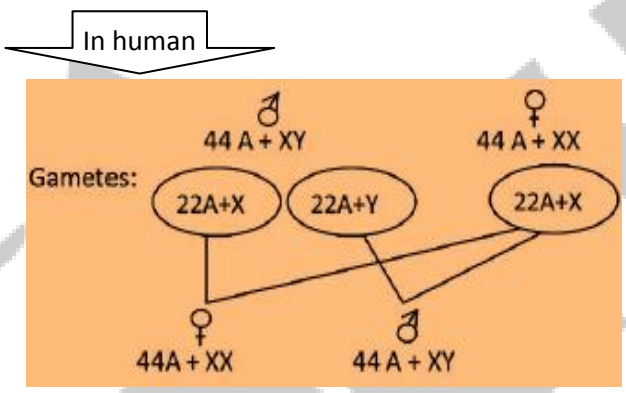




***b.* XX female –XY male mechanism:** Male is heterogametic (X & Y) and female is homogametic (X only).

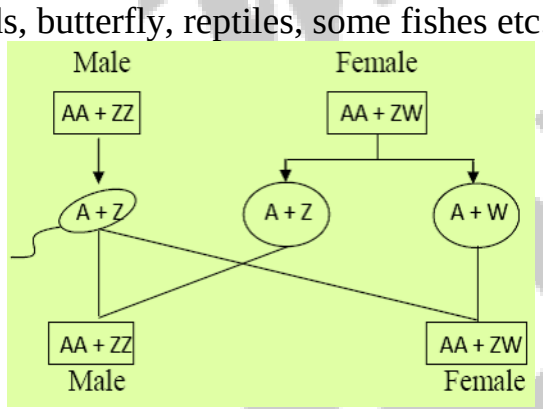
E.g. Human & *Drosophila.*





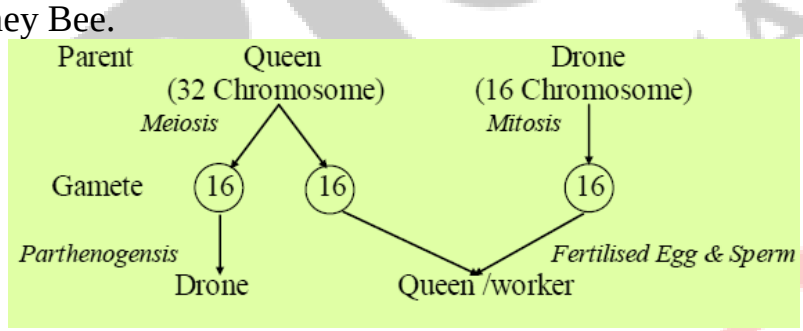
c. ZZ male -ZW female mechanism: Male is homogametic (with two Z) and female is heterogametic (Z & W).

E.g. Birds, butterfly, reptiles, some fishes etc.



d. Haplodiploid mechanism: It is based on the number of sets of chromosomes an individual receives.

E.g. Honey Bee.



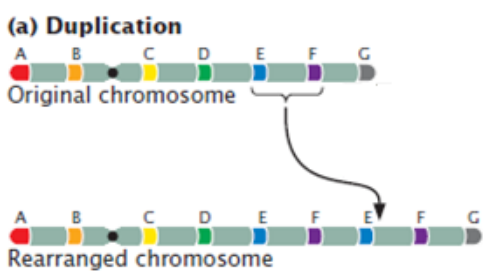
Mutation

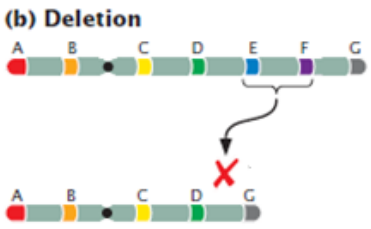
➔ *It is a sudden heritable change in DNA sequences resulting in changes in the genotype (and sometimes phenotype) of an organism.*

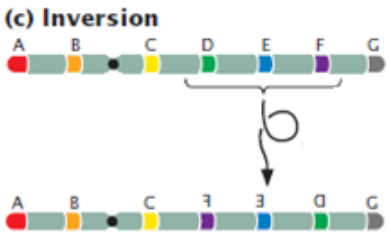
- **Mutagens:** Agents which induce mutation. 2 types-
  - **Physical mutagens:** UV radiation, X-ray, **α, β, γ** rays etc.
  - **Chemical mutagens:** Mustard gas, phenol, formalin etc.

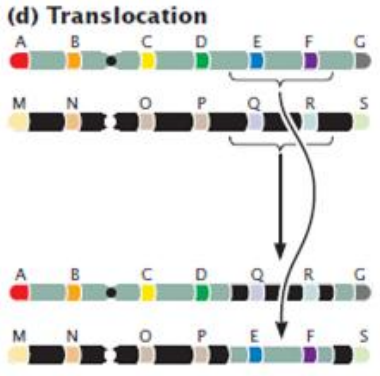
Types of Mutations-

- **Frame-shift mutation:** Loss (deletions) or gain (insertion/ duplication) of a DNA segment.
- **Point mutation**: Mutation due to change in a single basepair of DNA. E.g. Sickle cell anemia









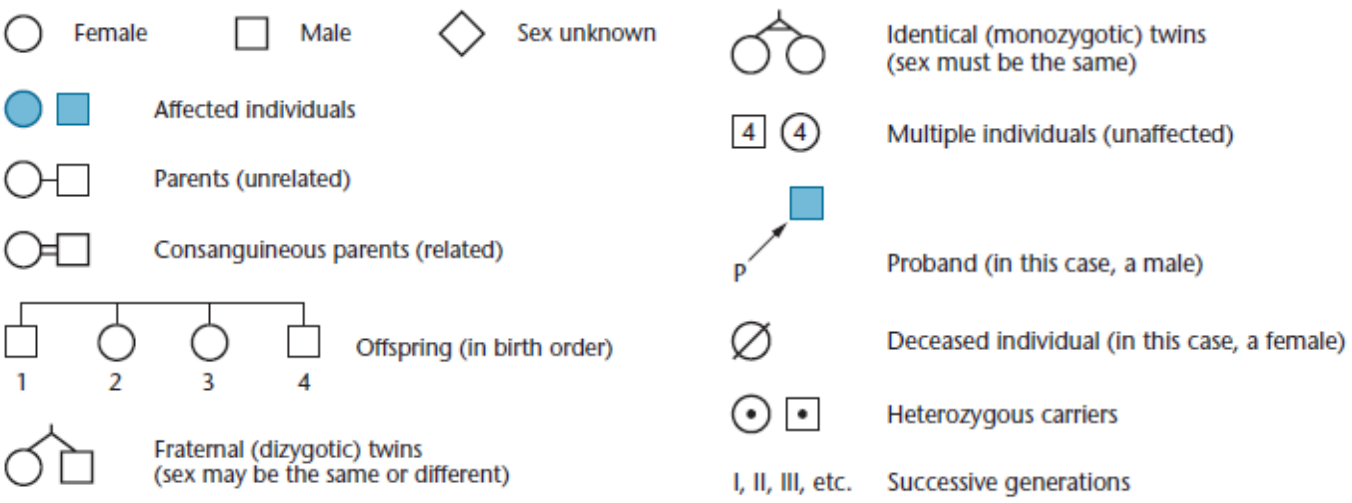
PEDIGREE ANALYSIS

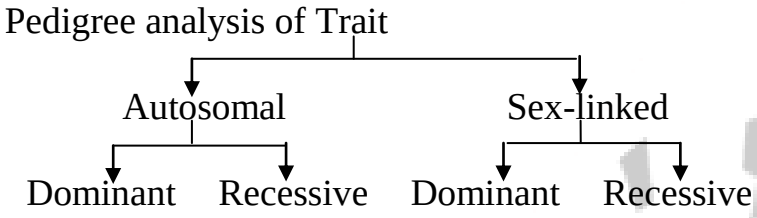
➔ *It is analysis of inheritance of a particular trait through several generations in a family.* The representation or chart showing family history is called **family tree (pedigree).**

This is a very useful method to study human genetics because:-

- Human beings have a long generation time.
- Produce extremely small no. of offspring which make human genetic studies difficult.
- In human, control crosses are not possible to study inheritance.

Symbols used in the human pedigree analysis:-



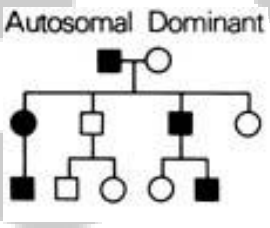


(a) Autosomal dominant trait

E.g. Myotonic dystrophy, polydactyly, tongue rolling ability etc.

Features:-

* Trait does not skip generations
* Affected individuals all have an affected parent.
* Appears equally in both sexes.

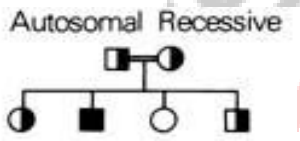


(b) Autosomal recessive trait

E.g. Sickle-cell anaemia, Albinism etc.

Features:-

* Appear only when homozygous, hence trait may skip generations.
* Appearance of affected children from normal parents (heterozygous). All children of affected parents are also affected.
* Appears equally in both sexes.



Genetic Disorders

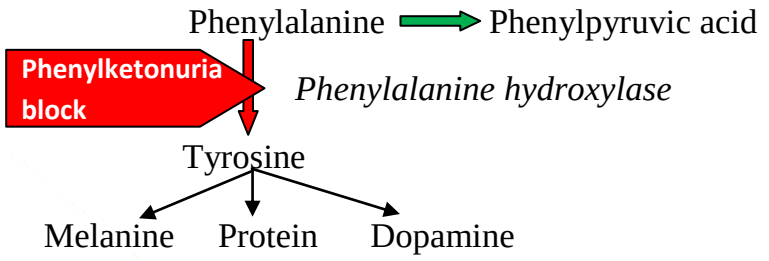
2 types: **Mendelian** disorders and **Chromosomal** disorders.

1. Mendelian Disorders

- *Caused by alteration or mutation in the single gene.*
- It may be dominant or recessive which can be traced by the pedigree analysis.

E.g. **Haemophilia, Cystic fibrosis, Sickle-cell anaemia, Colour blindness, Phenylketonuria, Thalesemia, etc.**

Mendelian Disorder	Symptoms	Cause	Features
Haemophilia (Royal disease)	A simple cut results in non-stop bleeding.	A protein involved in the blood clotting is affected.	• Sex linked recessive disease. • The heterozygous female (carrier) for haemophilia may transmit the disease to sons. • The possibility of a female becoming a haemophilic is very rare because mother has to be at least carrier and father should be haemophilic (unviable in the later stage of life). • **Queen Victoria** was a carrier of the disease. Her pedigree shows a number of haemophilic descendents.
Colour blindness	Failure to discriminate red and green colour.	Defect in certain genes present in the X chromosome.	• Sex-linked recessive disorder • It occurs in about 8% of males and only about 0.4% of females. This is because the genes are on the X chromosome. • The son of a woman who carries the gene has a 50% chance of being colour blind. A daughter will not normally be colour blind, unless her mother is a carrier and her father is colour blind.

Sickle-cell anaemia	- The mutant ***Hb*** undergoes polymerization under low O_2-tension causing the change in shape of the RBC to *sickle structure*. -RBCs are destroyed more rapidly leading to **anaemia**.	- The defect is caused by the substitution of **Glutamic acid (Glu)** by **Valine (Val)** at the **6th position** of the **β - globin chain** of the Hb. - This is due to the single base substitution at the 6th codon of the **β -globin gene** from **GAG** to **GUG.**	• Autosomal recessive trait. • It can be transmitted from parents to the offspring when both the partners are carrier (or heterozygous). • 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*
Phenyl-ketonuria	**- Mental retardation** **- Reduction in hair** **- Skin pigmentation**. **-** ***Phenyl pyruvic acid*** excreted through urine because of poor absorption by kidney.	- The disease is caused by mutation in the gene that code for the enzyme ***phenylalanine hydroxylase*** that converts the amino acid ***phenylalanine*** into ***tyrosine.*** As a result, phenylalanine accumulates and converts into ***phenyl pyruvic acid*** which accumulate in brain.	• Autosomal recessive disorder (single gene mutation). • An inborn error of metabolism. 
Thalassemia	Anaemia	- Due to either mutation or deletion which ultimately results in reduced rate of synthesis of one of the globin chains (α and β chains) that leads to formation of **abnormal Hb**.	• Autosomal recessive disorder • Blood disease transmitted from parents to the offspring when both the partners are carrier (or heterozygous). • 2 types- α and β ➢ In **α Thalassemia**, production of α globin chain is affected. It is controlled by genes HBA1 and HBA2 on chromosome 16 of each parent and it is observed due to mutation or deletion of one or more of the 4 genes. ➢ In **β Thalassemia**, production of β globin chain is affected. It is controlled by a single gene HBB on chromosome 11 of each parent and occurs due to mutation of one or both the genes.

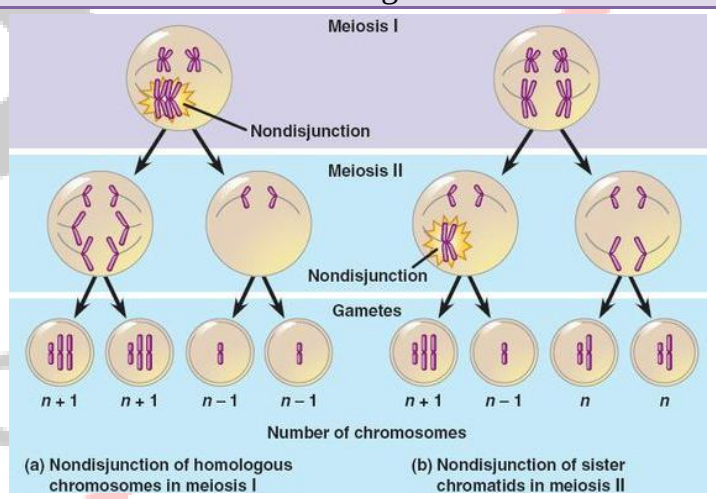
2. Chromosomal disorders

- *They are caused due to absence or excess or abnormal arrangement of one or more chromosomes. 2 types:*

a. Aneuploidy: The gain or loss in a subset of chromosomes due to failure of segregation of chromatids during cell division.

It includes,

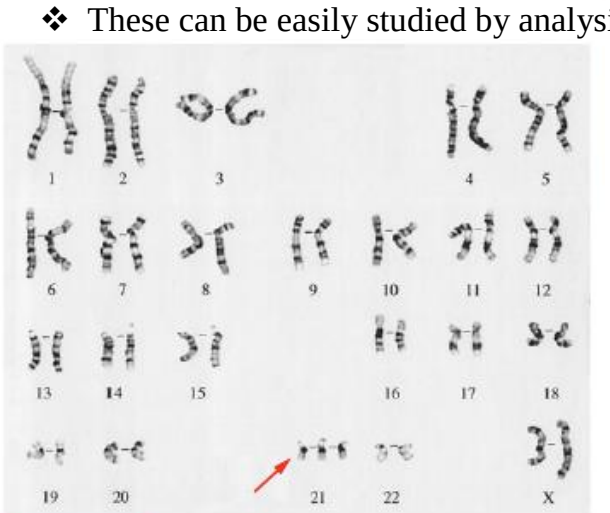
- **Nullysomy (2n-2):** A chromosome pair is lost from diploid set.
- **Monosomy (2n-1):** One chromosome is lost from diploid set.
- **Trisomy (2n+1):** One chromosome is added to diploid set.
- **Tetrasomy (2n+2):** 2 chromosomes are added to diploid set.



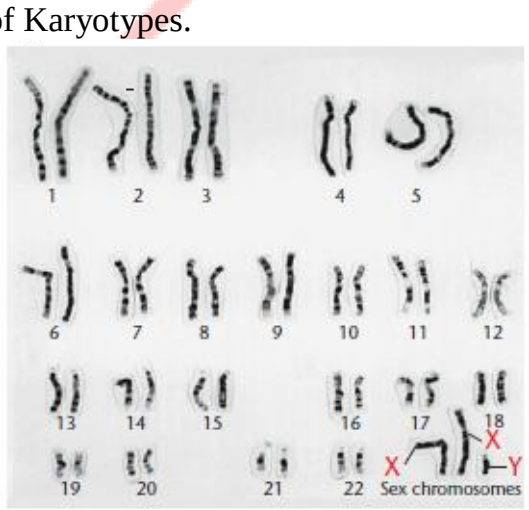
Examples for chromosomal disorders

Chromosomal disorder	Karyotype/ Genetic constitution	Phenotype
Down's syndrome	**45 A + XX / 45 A + XY** (trisomy of chromosome 21)	♥ Mental retardation. ♥ Short statured with small round head & broad flat face ♥ Partially open mouth with big & wrinkled tongue. ♥ Broad palm with palm crease. Many loops on finger tips. ♥ Heart defects.
Klinefelter's syndrome	**44 A + XXY** (trisomy of X-chromosome in **male**)	♥ Sterile male (immature testes & sex glands) ♥ Mentally retarded ♥ Overall masculine development ♥ Feminine characters (development of breast, i.e., **Gynaecomastia**) is also expressed
Turner's syndrome	**44 A + XO** (Monosomy of chromosome in **female**)	♥ Sterile (Poorly developed ovaries & sex glands) ♥ Lack of other 2^0 sexual characters

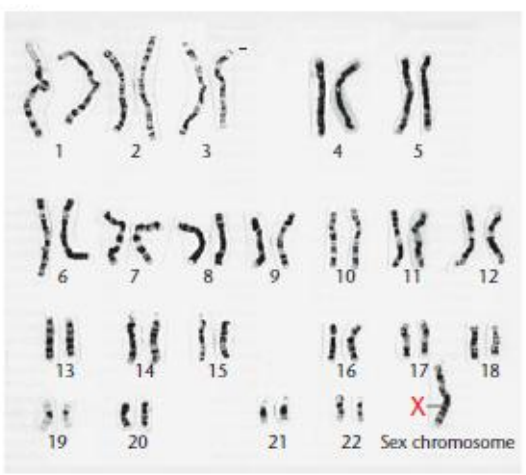
❖ These can be easily studied by analysis of Karyotypes.



Down's syndrome



Klinefelter's syndrome



Turner's syndrome

b. Polyploidy (Euploidy): It is an increase in a ***whole set of chromosomes*** due to failure of cytokinesis after telophase stage of cell division. This is often seen in plants.