

RIZVI COLLEGE OF ARTS SCIENCE AND COMMERCE

Chap 3 – Inheritance and Variation

Class : **SYJC Science**

Mrs. Madhuri Mane
Sr. Teacher (Biology Department)
Junior college

INHERITANCE AND VARIATION

Total Marks – 06(04)

BY:-- MRS. MADHURI MANE

CHROMOSOMES AND MECHANISM OF INHERITANCE

Heridity/ Inheritance

Transmission of genetic information from generation to generation.

Gregor Mendel (Moravia--1822)

- *Gave accurate explanation for the mechanism of inheritance by using hybridization technique.*
- *Studied seven traits in garden pea. (*Pisum sativum*) one at a time.*
- *Mendel postulated the principles of heridity which later became the fundamental laws of heridity. Proposed by Correns(1900)*

→ Mendel visualised, traits not inherited physically but by 'something' present inside the gametic cell.

→ Something ---- **factor**

→ Mendelian factor now termed as '**Gene**'.

→ Factors occurs in pairs in Parents and segregate from each other during gamete formation.



Gregor Johann
Mendel(1822--1884)

SUCCESS.

1. Experiments were **carefully planned** and involved **large sample**.
2. Carefully recorded the number of plants and **expressed results as ratios**.
3. Pea plant, **contrasting characters** can be **easily recognized**.
4. **Seven different characters** in pea plant were controlled by a **single factor**.
5. The **factors** are located on **separate chromosomes** and are **transmitted** from **generation to generation**.
6. Introduced the concept of **dominance** and **recessiveness**.

Mendel studies seven characteristics in the garden pea

	Flower color	Flower position	Seed color	Seed shape	Pod shape	Pod color	Stem length
P	Purple	Axial	Yellow	Round	Inflated	Green	Tall
	White	Terminal	Green	Wrinkled	Constricted	Yellow	Dwarf
F ₁	Purple	Axial	Yellow	Round	Inflated	Green	Tall



Genetic Terminology

1. **Character** – It is feature of organism. Eg. Height of stem.
2. **Trait** – An inherited character and its detectable variant. Eg. Tall or Dwarf.
3. **Factor** – It is unit of heredity, a particle present in the organisms which is responsible which is responsible for the inheritance and expression of character.
4. **Gene** – It is a particular segment of DNA which is responsible for the inheritance and expression of that character.(Term- Johannsen)
5. **Allels of Allelomorphs** – The two or more alternative forms of gene are called alleles of each other. They occupy identical loci on homologous chromosomes.(Bateson)

6. **Dominant** – It is an allele that expresses its trait even in the presence of an alternative allele.
7. **Recessive** – It is an allele which is not expressed in the presence of an alternative allele.
8. **Phenotype** – The external appearance of an individual for any trait is called phenotype.
9. **Genotype** – It is representation of the genetic constitution of an individual with respect to a single character or set of characters.
10. **Homozygous** – An individual possessing similar alleles for a particular trait is called homozygous or pure for that trait.
11. **Heterozygous** – An individual possessing dissimilar alleles for a particular trait is called heterozygous or hybrid for that trait.

12. **Pure line** – An individual or a group of individual that is homozygous or true breeding for one or more traits.
13. **Monohybrid** – It is heterozygous for one trait and produced in a cross between two pure parents differing in single pair of contrasting characters.
14. **F₁ generation** – The progeny produced from a cross is called first filial of F₁ generation.
15. **F₂ generation** – The second generation produced by selfing of F₁ generation offsprings is called second filial or F₂ generation.
18. **Punnett** – Probability table representing different permutations and combinations of fertilization between gametes of opposite sex.
19. **Homologous chromosomes** – The morphologically, genetically and structurally essentially identical chromosomes present in a diploid cell are called homologous chromosomes .

20. Back cross – Cross of F₁ progeny with any of the parents.
21. Test cross – Cross of F₁ progeny with homozygous recessive parent.
22. Phenotypic ratio – Ratio of the offsprings produced in F₂ with respect to their physical appearance.

3Tall : 1Dwarf

23. Genotypic ratio – Ratio of the offsprings produced in the F₂ generation with respect to their genetic makeup.

1TT : 2Tt : 1tt

MONOHYBRID CROSS

Definition

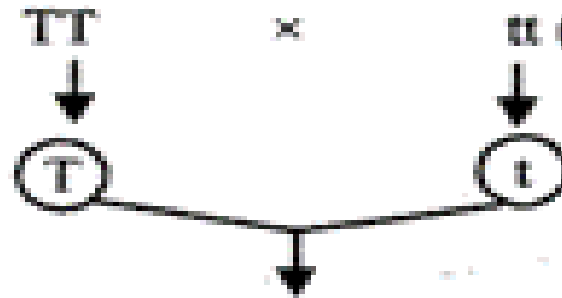
- A cross between parents differing in only one heritable trait.

- Mendel crossed **tall** (female) **TT** pea plant with **dwarf**(male) **tt** pea plant.
- In F_1 generation he gets all tall plants. (**Tt**)
- He allowed the **selfing of F_1** to get **F_2 generation**

Monohybrid cross

Parents : (Pure tall) TT \times tt (Dwarf)

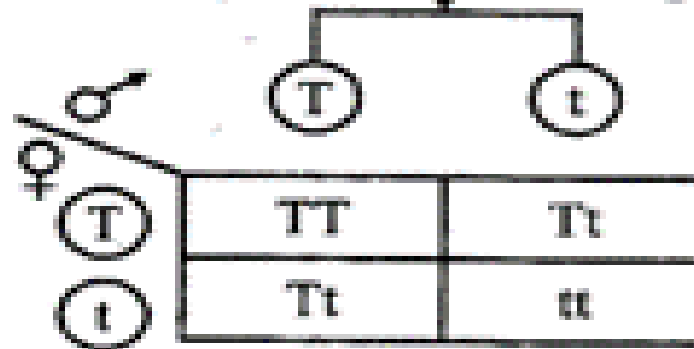
Gametes :



F_1 generation :

Tt Heterozygous tall
selfing

F_2 generation :



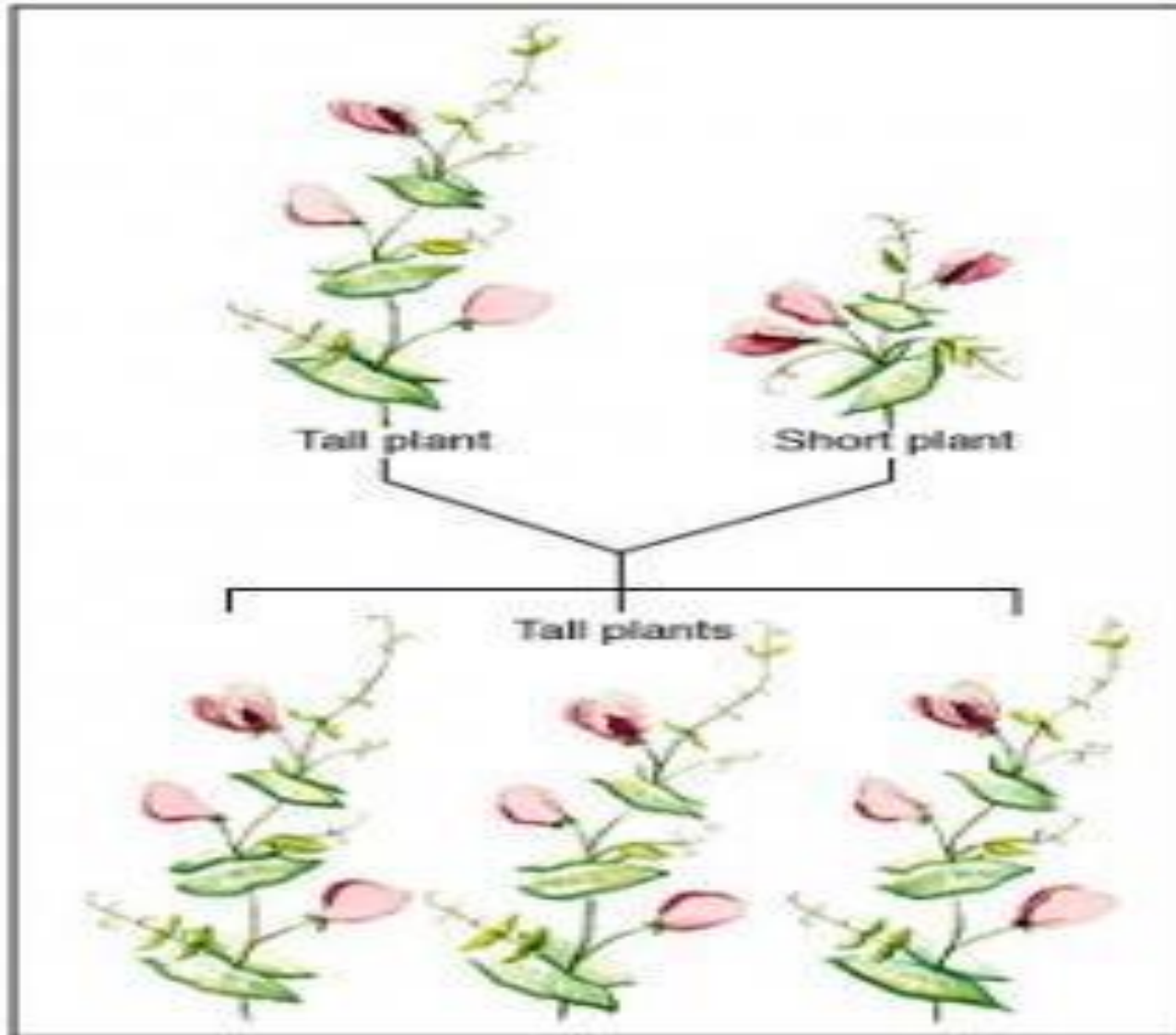
Phenotype ratio : 3 tall : 1 dwarf

Genotype ratio : Pure tall : hybrid tall : dwarf

1 : 2 : 1

Monohybrid cross

F1 generation all Tall



Monohybrid Cross

Parental Generation



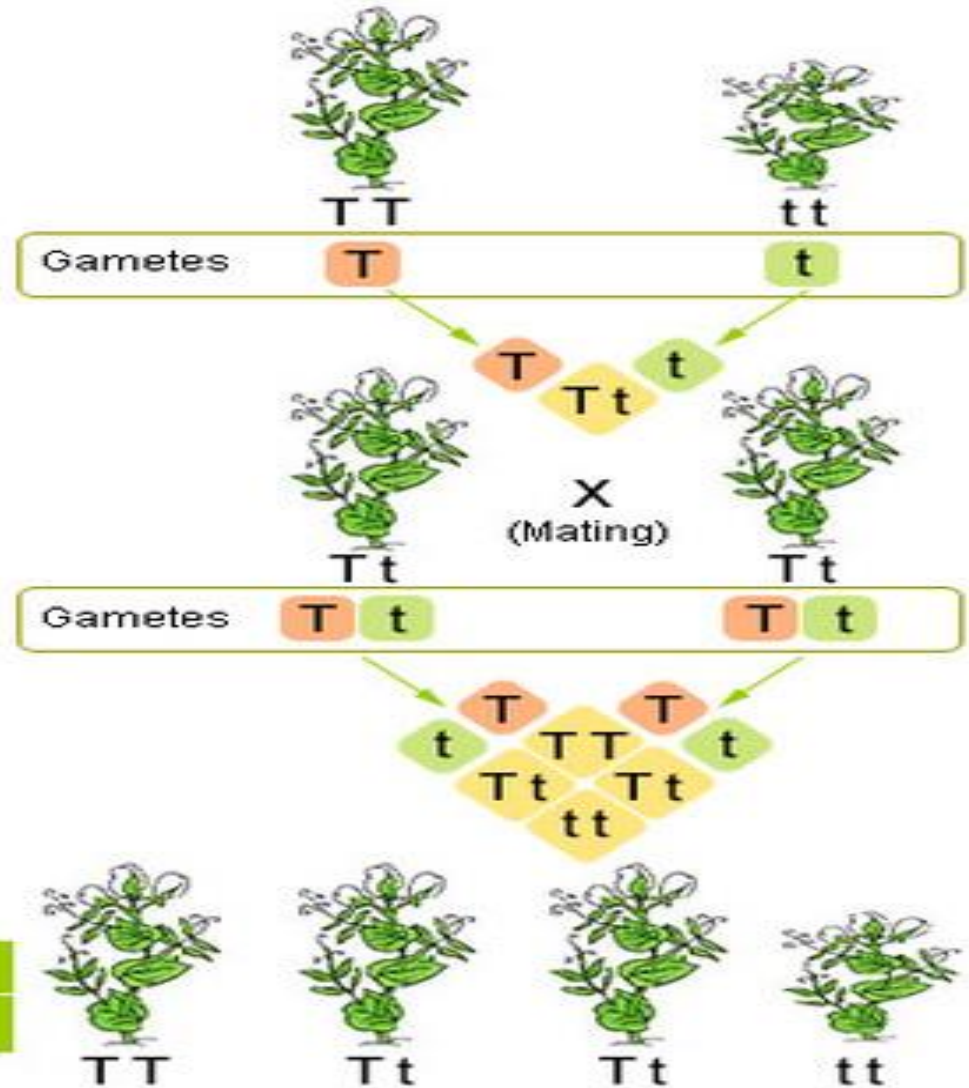
F1 Generation

Genotype	All are Tt.
Phenotype	All are tall



F2 Generation

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



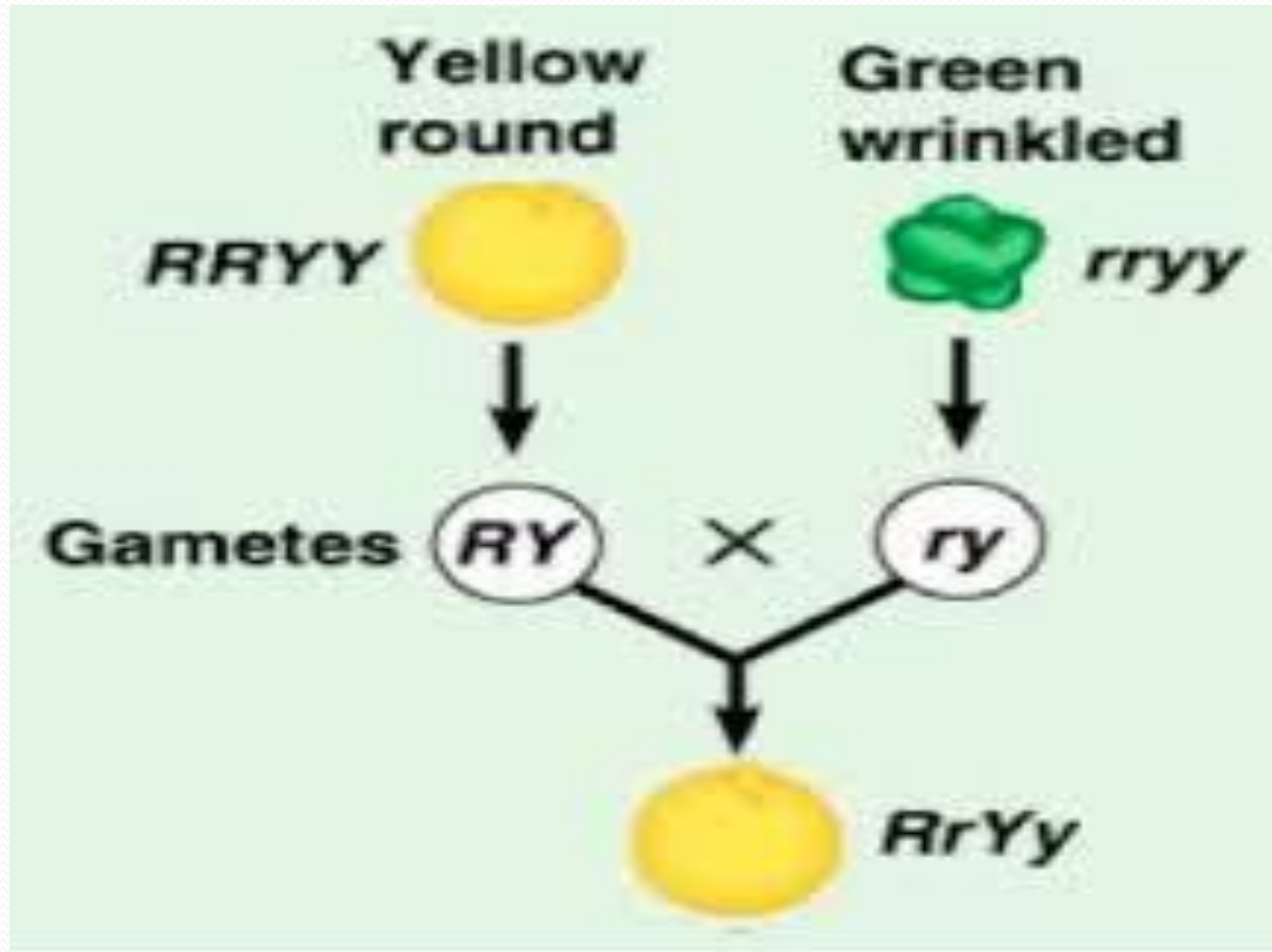
DIHYBRID CROSS

Definition

A cross between parents differing in two heritable traits.

1. Mendel crossed pea plants with **yellow and round** seeds as *female* plant & **green and wrinkled** seed as *male* plant.
2. F_1 generation showed all plants with **yellow round** seeds. (Yellow & round dominant)
3. Selfing of F_1 generation is done to get F_2 generation.

Dihybrid cross



Dihybrid cross

P

true-breeding
yellow, round peas

YYRR

x

yyrr

true-breeding
green, wrinkled peas

y = green
r = wrinkled

Y = yellow
R = round

F₁

generation
(hybrids)

yellow, round peas

All HYBRIDS!

100%



YyRr

self-pollinate

F₂

generation

Dominant/
Dominant

Recessive
/Dominant

Dominant/
Recessive

Recessive/
Recessive

9/16
yellow
round
peas

3/16
green
round
peas

3/16
yellow
wrinkled
peas

1/16
green
wrinkled
peas

Mendel's
Magic
Ratio of
Hybrids!

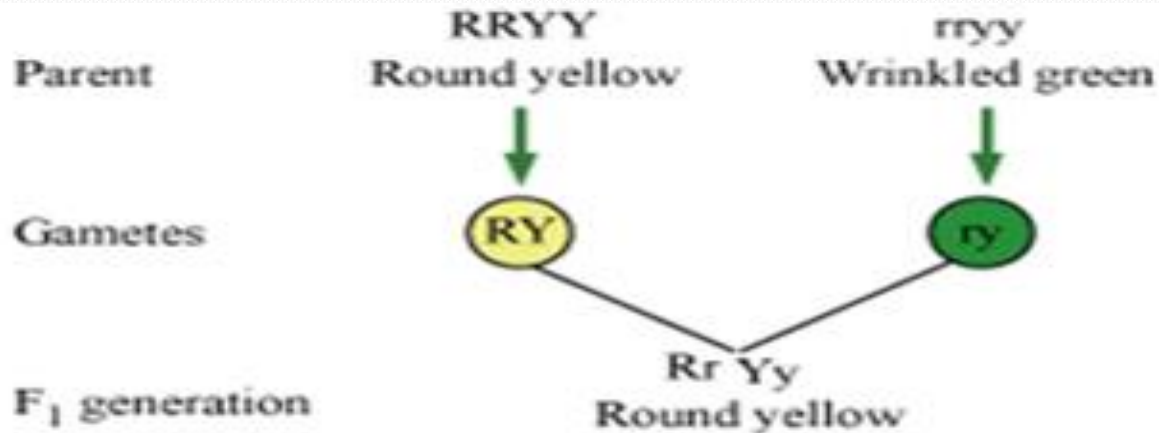
9:3:3:1



Dihybrid Cross – selfing of F1 generation

♀ \ ♂	YR	Yr	yR	yr
YR	 YYRR	 YYRr	 YyRR	 YyRr
Yr	 YYRr	 YYrr	 YyRr	 Yyrr
yR	 YyRR	 YyRr	 yyRR	 yyRr
yr	 YyRr	 Yyrr	 yyRr	 yyrr

Dihybrid Cross



	Ry	ry	Ry	ry
Ry	RRYY Round yellow	RrYY Round yellow	RRYy Round yellow	RrYy Round yellow
ry	RrYY Round yellow	rrYY Wrinkled yellow	RrYy Round yellow	rrYy Wrinkled yellow
Ry	RRYy Round yellow	RrYy Round yellow	RRyy Round green	Rryy Round green
ry	RrYy Round yellow	rrYy Wrinkled yellow	Rryy Round green	rryy Wrinkled green

INHERITANCE

Law of Dominance

Explain the expression of only one of the parental characters of a monohybrid cross in F₁.

Statement of Law of Dominance.

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

Law of segregation (Law of Purity of gametes)

Alleles do not show any mixing and both the alleles are recovered as such in F₂ generation though one of these is not seen at f₁ stage.

Statement of law of Segregation.

When hybrid (F₁) forms gametes, the alleles segregate from each other and enter in different gametes.

The gametes form are pure in that they carry **only one allele each**, hence also called Law of Purity of gametes.

Law of Independent Assortment.

Genes or alleles present on separate chromosomes independently separate from each other, during formation of gametes and then randomly unite in fertilization.

Statement of Law of Independent Assortment

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

BACK CROSS

F_1 individuals obtained is **crossed** with **one of the two parents** from which they were derived .

(either dominant or recessive)

Test Cross

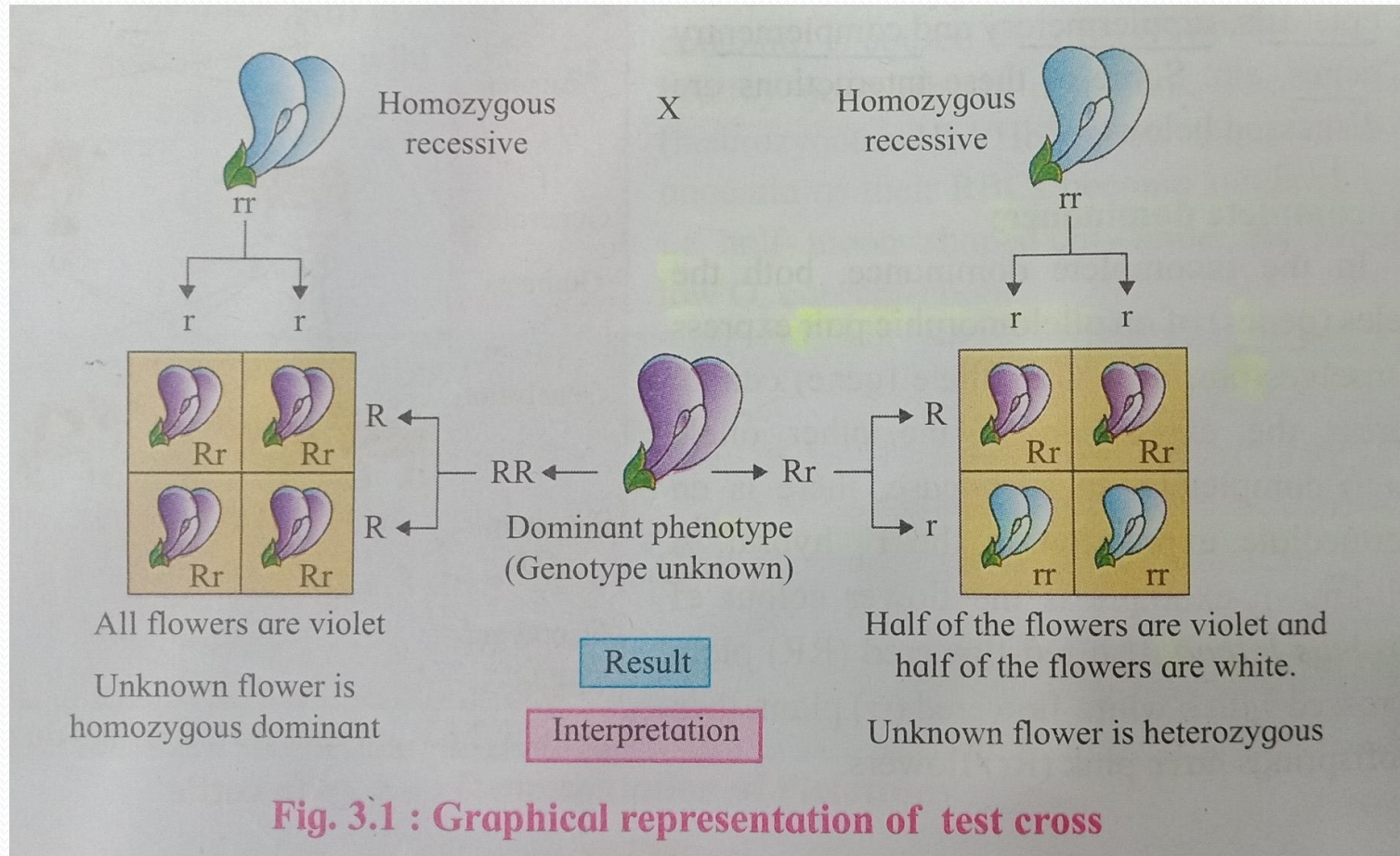
Cross of F₁ hybrid with the homozygous recessive parent is known as a test cross.

It is used to test whether an individual is homozygous or heterozygous.

Eg. A pea plant having **violet flowers** is crossed with a pea plant with **white flowers**.

- If all flowers produced are **violet** – Plant is **pure homozygous**.
- If we get **violet** and **white flowers**(1:1) – Plant is **heterozygous**.

Test Cross



Generalizations by Mendel, on the basis of his experiments.....

1. *Single trait (Height) → Single gene → Two alleles (Tt).*
2. *Two alleles show interaction in which one is completely dominant. (Tt)*
3. *Factors (genes) for different traits present on different chromosomes assort independently.*



Do you know ?

The deviations are :

- a. Single trait \longrightarrow Single gene \longrightarrow two alleles showing interactions like codominance and incomplete dominance.
- b. Single trait \longrightarrow Single gene \longrightarrow more than two alleles (multiple allelism).
- c. Single trait \longrightarrow more than one genes (Polygenic inheritance) showing different epistatic interactions or additive effect.
- d. Single gene influencing many traits (pleiotropy).

DEVIATIONS FROM MENDELIAN RATIOS

Post-Mendelian genetics or Neo Mendelian genetics

Gene interactions :

1. Intragenic interactions . (Allels of same gene)
 - a) Incomplete dominance
 - b) Co-dominance
 - c) Multiple alle
2. Intergenic interactions (Allels of different genes
on same or different chromosome)
 - a) Pleiotropy
 - b) Polygenes
 - c) Epistasis
 - d) Supplementary and complementary genes.

A. Incomplete Dominance

- Both the alleles(genes) of an allelomorphic pair express themselves partially.
- One allele(gene) cannot suppress the expression of the other allele completely.
- There is an intermediate expression in the F1 hybrid.
- *Eg. Mirabilis jalapa*
- If the Red-flower(RR) plant is crossed with a white-flower(rr) plant, then F1 offsprings have pink(Rr) flowers.

Incomplete Dominance

P generation



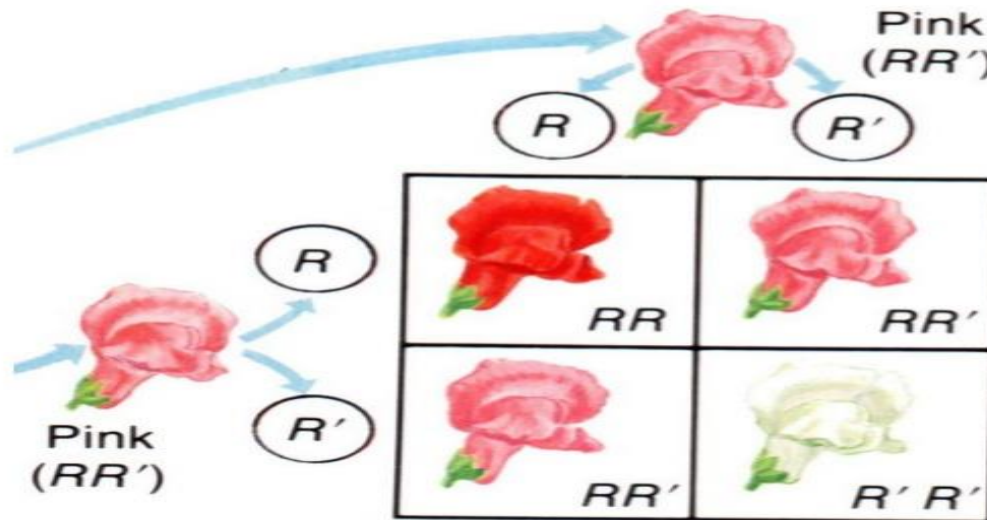
Gametes



F₁ generation



Incomplete Dominance- F₂ generation



B. Co-Dominance

- Both the alleles (genes) of an allelomorphic pair express themselves equally in F1 hybrids.
- Alleles which are able to express themselves equally independently in hybrids, are called co-dominant alleles.
- Both the alleles are expressed equally.
- Eg. Coat colour in cattle.
- When Red coat cattle (RR) are crossed with white coat colour (WW), F1 hybrids (RW) are Roan.

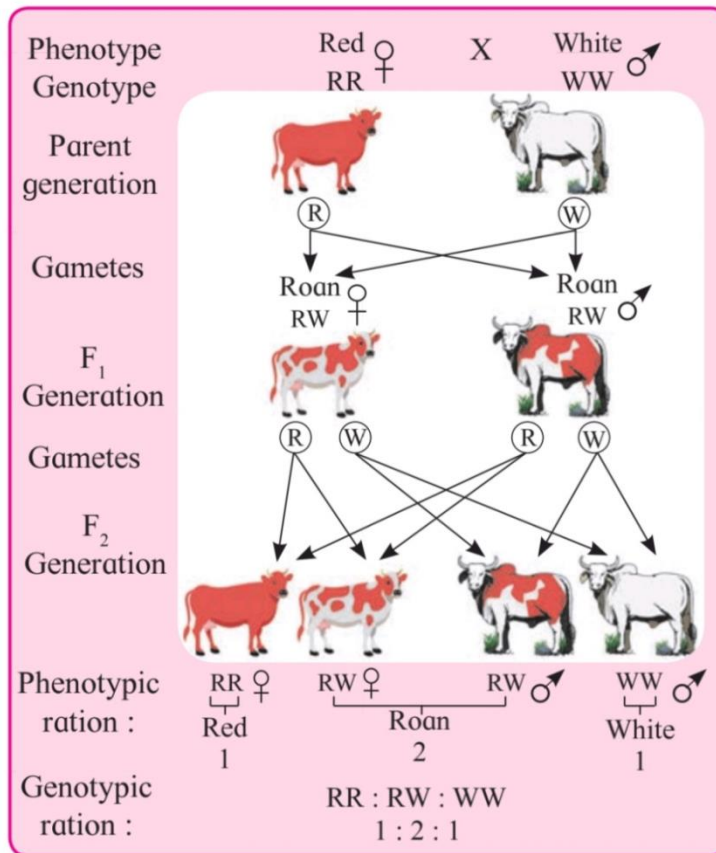







Fig. 3.2 : Representation of co-dominance

C. Multiple alleles

- **More than two alternative forms (alleles)** of a gene in a population occupying the same locus on a chromosome or its homologue.
- It arise by **mutations** of the **wild type** of gene.
- Wild type is **dominant** over all other mutant alleles.
- In **Drosophila**, a large number of multiple alleles are known, eg. *Size of wings from normal wings to vestigial wings. Normal wing is wild type while vestigial is recessive type.*

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

D. Pleiotropy

- *When a single gene controls two or more different traits it is called pleiotropic gene and the phenomenon is called pleiotropy.*
- *Phenotypic ratio is **1:2** instead of **3:1** due to death of recessive homozygote*
- $Hb^A \rightarrow$ **Dominant healthy gene**
- $Hb^s \rightarrow$ **Sickle-cell anaemia**
- $Hb^A/Hb^s \rightarrow$ **carriers - mild anaemia**

(RBC becomes sickle or half moon shaped)
due to low O_2 concentration.

PARENTS

Phenotype

Sickle-cell carrier

×

Sickle-cell carrier

Genotype

Hb^A Hb^S

Hb^A Hb^S

GAMETES

Hb^A

Hb^S

Hb^A

Hb^S

Genotype

Hb^A Hb^A

Hb^A Hb^S

Hb^A Hb^S

Hb^S Hb^S

OFFSPRINGS

Phenotypes

Normal

Sickle-cell carrier

Sickle-cell anaemic

1

2

1 (dies)

Fig. 3.4 : Representation of Pleiotropy

Gregor Johann Mendel – published inheritance – 1866, remained unnoticed till 1900.

His approach of using maths and statistics to explain biological phenomenon was totally new and unacceptable to then biologists.

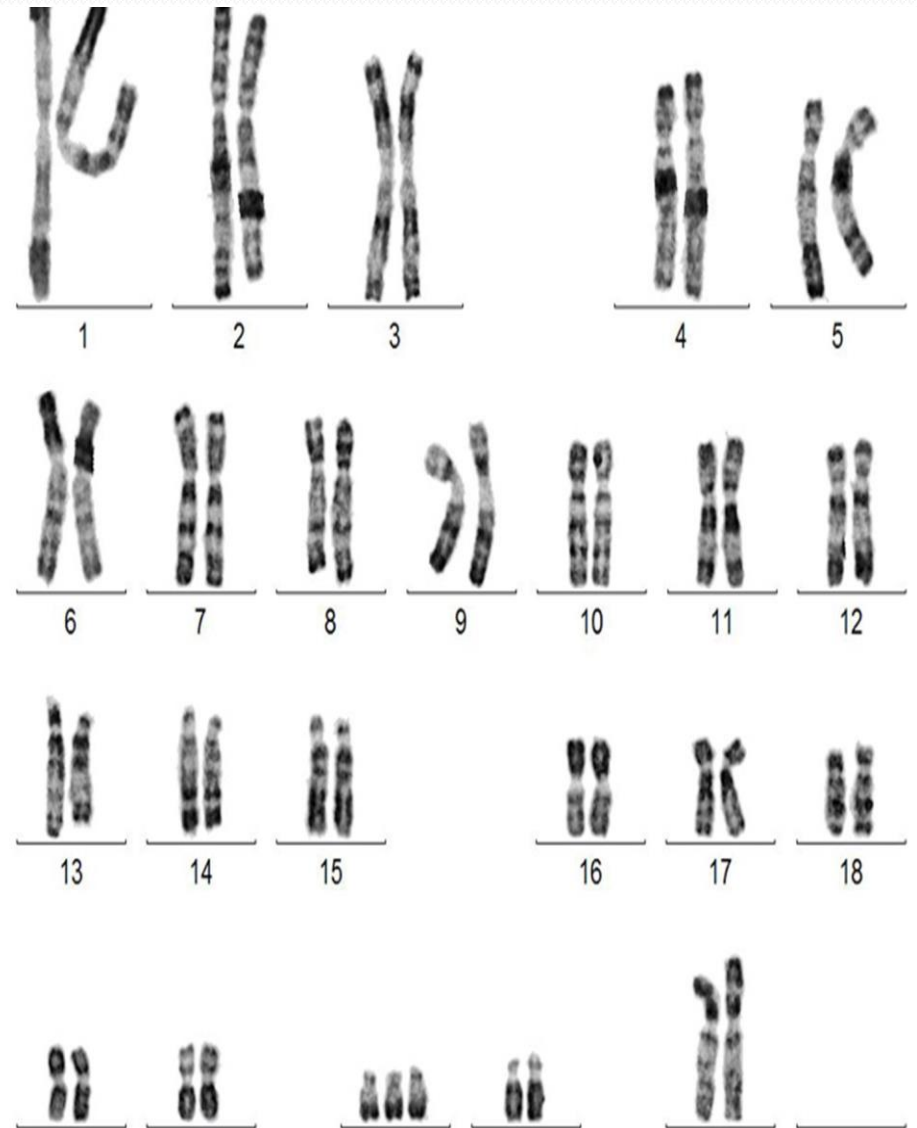
In 1900, three scientist Hugo de vries, correns and von rediscovered Mendel's work.

Based on these observation, chromosomal theory of inheritance was put forth by Sutton and Boveri.

CHROMOSOMAL THEORY OF INHERITANCE

- Chromosomes are present in **pairs** in **somatic cells**.
- During **gamete formation** homologous chromosomes pair, **segregate** and assort independently during **meiosis**.
- Each **gamete** contains only **one chromosome** from a **pair**.
- **Nucleus** of gametes (male ♂ female) contains chromosomes, which carry all **hereditary traits**.
- **Fusion** of haploid male gamete and female gamete **restores** the **diploid number** of **chromosomes** of the species.

Human karyotype



CHROMOSOMES

1. **Filamentous bodies** in eukaryotic nucleus.
2. Chromo = **colour**, soma = **body**.
3. Size metaphase chromosome – **0.1 to 33 micrometer** in length and **0.2 to 2 micrometer** in thickness.
4. Visible during **cell division**.
5. Capable of **self-replication**.
6. Role – **heredity, mutation, variation** and **evolutionary development**.
7. Made – **DNA, histone** and **non-histone protein**.

Function – Chromosomes are carrier of heredity.

NUMBER OF CHROMOSOMES:

Ploidy – Degree of repetition of the primary basic number of chromosomes in cell.(x)

A. Euploidy – Chromosome number in a cell is the exact multiple of the primary basic number.

- *Monoploid/haploid* – One set of chromosomes.($x=n$)
- *Diploids* – Two sets of chromosomes.($2n$)
- *Triploids* – Three sets of chromosomes.($3n$)
- *Tetraploids* – four sets of chromosomes.($4n$)

B. Aneuploidy – Chromosome number is not the exact multiple of the haploid set.

It is either *addition* or *deletion* of one or more chromosome to the total number of chromosomes in a cell.

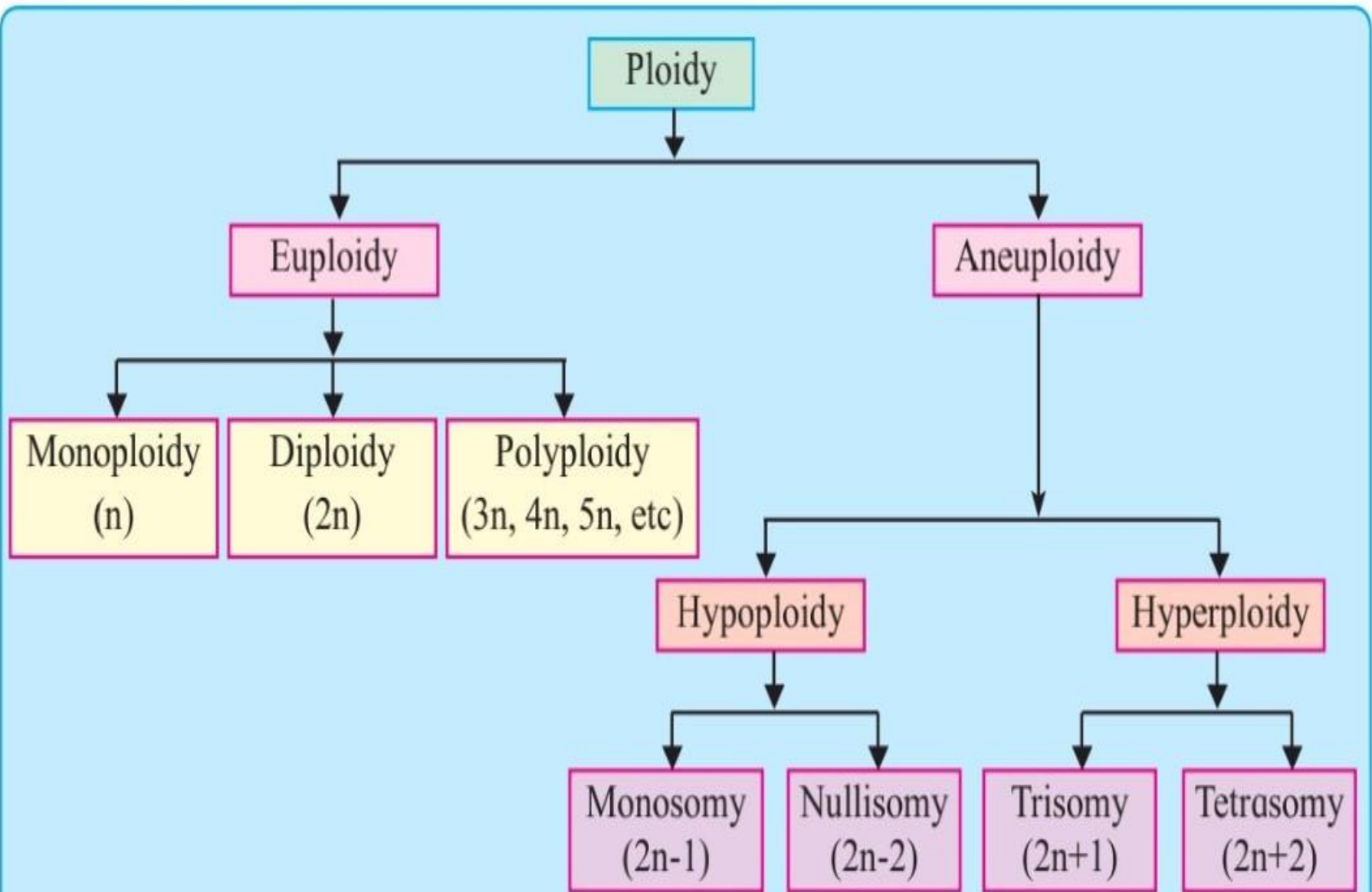
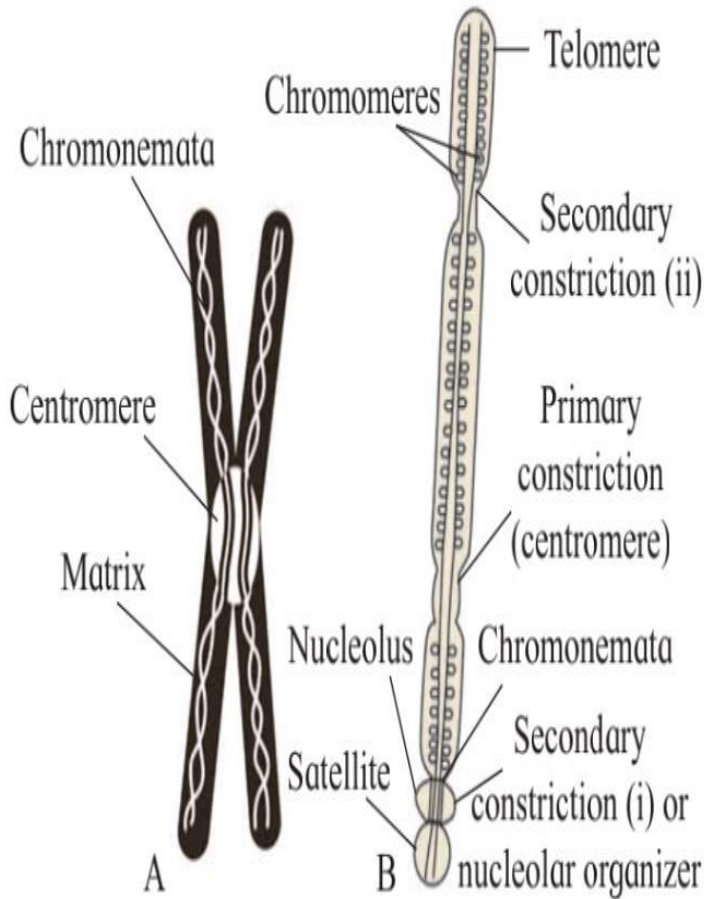


Chart 3.5 : Variation in chromosome number (ploidy)

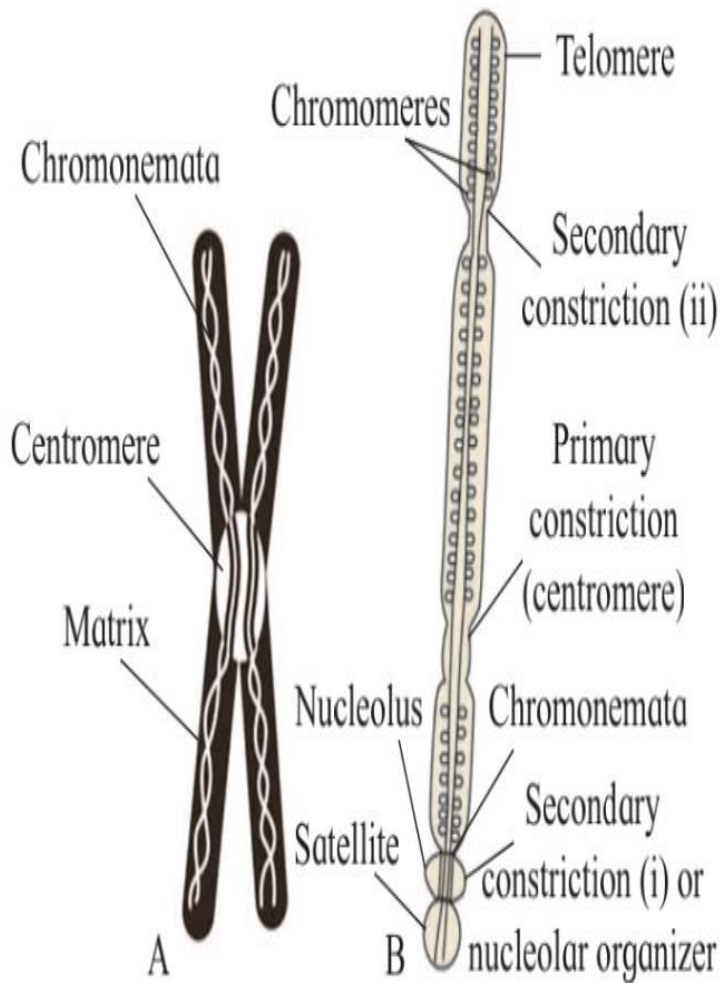
Structure of Chromosome.



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

Fig. 3.6 : Structure of Chromosome

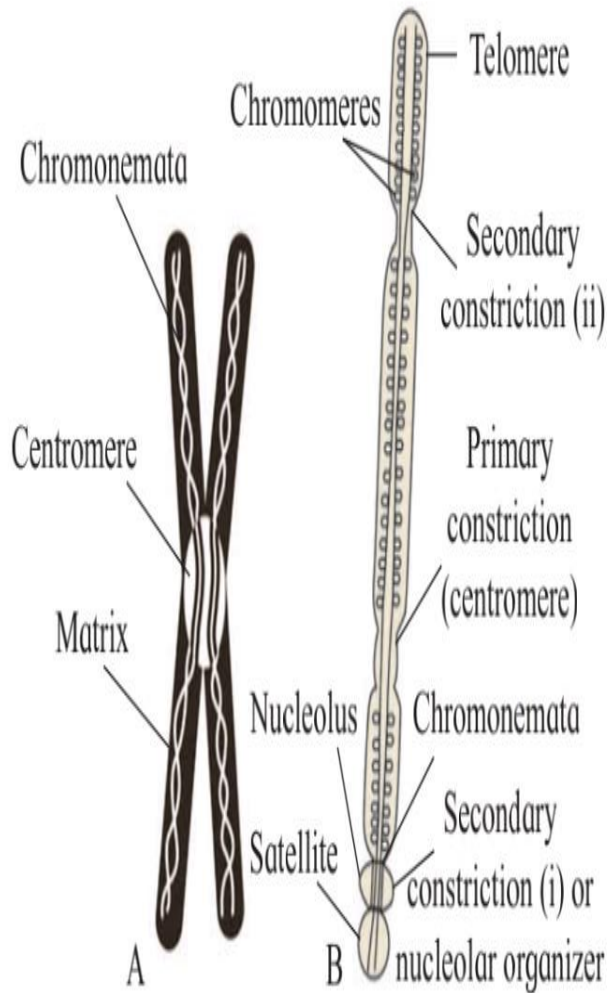
1. *Best visible – microscope – metaphase stage.*
2. *This stage – chromosomes are highly condensed.*
3. *Typical chromosome consists of two chromatids joined together at centromere or primary constriction.*



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

Fig. 3.6 : Structure of Chromosome

4. Primary constriction consists of a *disk* shape plate called *kinetochore*.
5. At kinetochore, *spindle fibres* get attached during cell division.
6. Some chromosomes possess additional *secondary constriction*.
7. At *sec. constriction I*, *nucleolus* becomes organized during *interphase*.



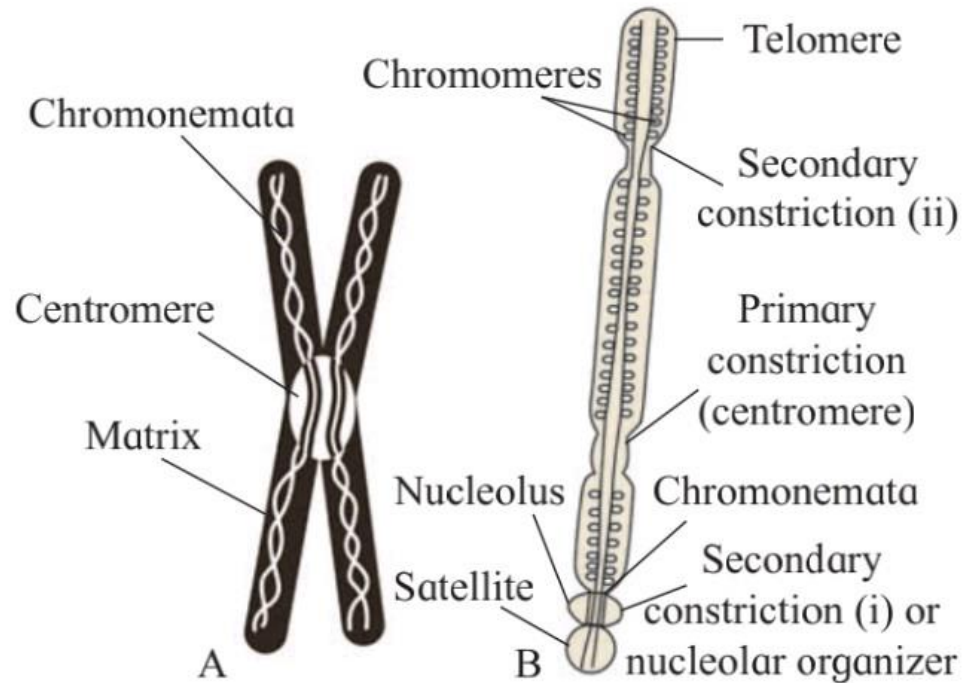
A : Parts of chromosomes B : Showing secondary constrictions and details

Fig. 3.6 : Structure of Chromosome

8. A **satellite body (SAT body)** is attached at **secondary constriction II** in few chromosomes.

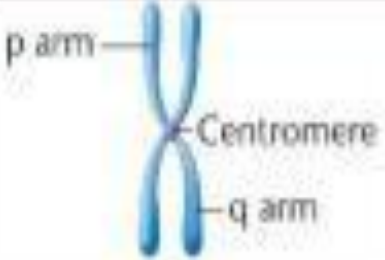
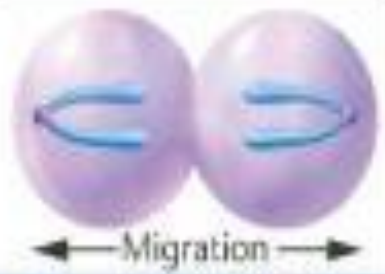






9. Chromatids in turn contain a **long, unbranched, slender, highly coiled DNA thread**, called **chromonema**, through its length.

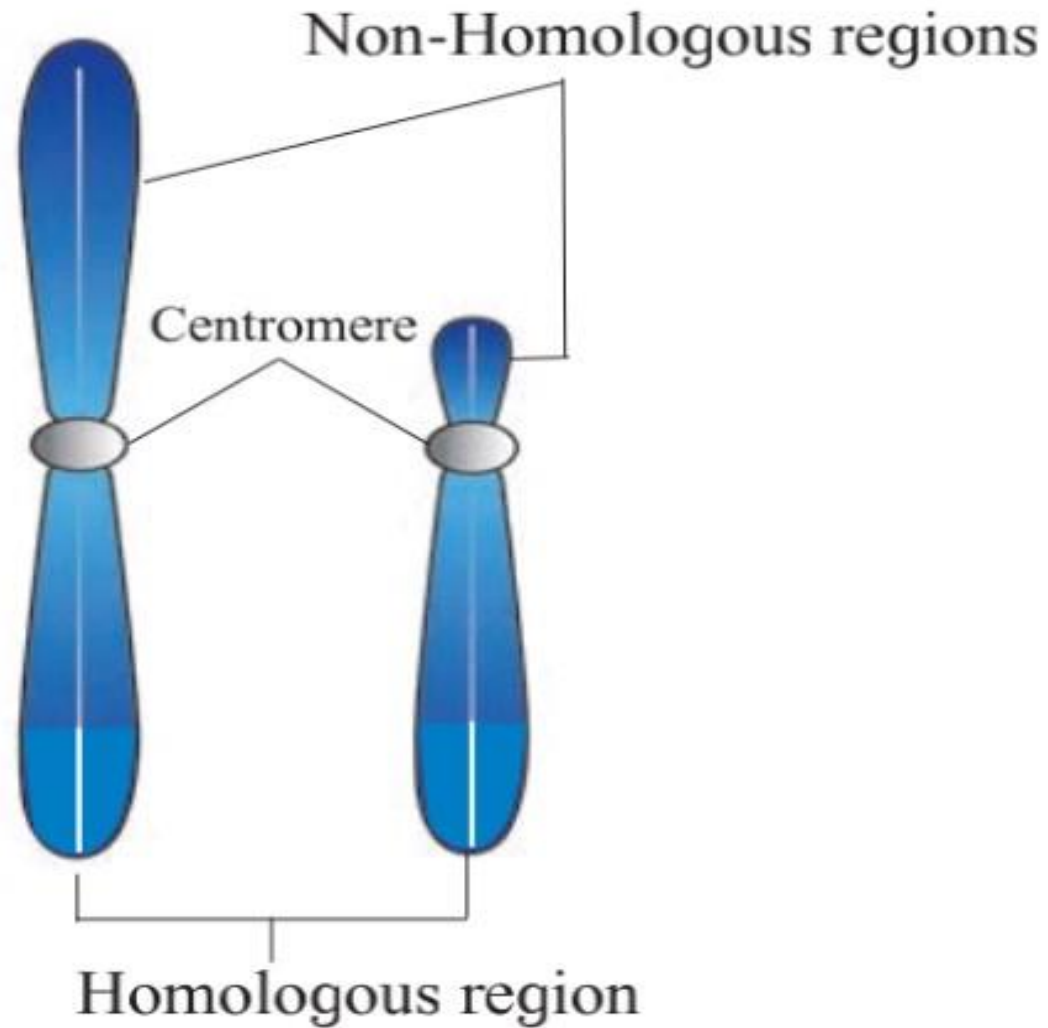
10. Chromatids consists of **double stranded DNA** molecule which extends from one end of chromosome to other.



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

Fig. 3.6 : Structure of Chromosome

Centromere location	Designation	Metaphase shape	Anaphase shape
Middle	Metacentric		
Between middle and end	Submetacentric		
Close to end	Acrocentric		
At end	Telocentric		



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

SEX CHROMOSOMES

- Sex Chromosomes are responsible for the **determination of sex**. (Allosomes)
- Human being/other mammals – **X & Y** sex chromosomes.
- Both show **Homologous** and **Non-Homologous** regions.
- Homologous region-- Show **similar genes**.
- Non-homologous region – **dissimilar genes**.
- **Crossing over** – Occurs between homologous regions of **X and Y chromosomes**.

Differentiate between

X chromosomes

1. It is straight and longer.
2. X chromosome is metacentric.
3. Large amount of euchromatin and small amount of heterochromatin.
4. Euchromatin has large amount of DNA, hence active.
5. X linked genes present.
6. Non-homologous region large.

Y chromosomes

1. It is bend and small.
2. Y chromosome is acrocentric.
3. Small amount of euchromatin and large amount of heterochromatin.
4. Euchromatin has less amount DNA, hence less active.
5. Y linked genes present.
6. Non-homologous region small.



LINKAGE

AND

CROSSING OVER

Linkage (Bateson-Morgan)

Definition

Tendency of two or more genes present on the same chromosomes that are inherited together are known as linkage.

Two types:--

- Complete linkage
- Incomplete linkage

Complete Linkage

*Linked genes which are closely located on the chromosomes do not separate and inherit together are called **completely linked genes** and their phenomenon of their inheritance is called **complete linkage**.*

→ Parental traits are inherited in offsprings.

→ Eg. X chromosomes of Drosophila males- shows complete *linkage*.

Incomplete linkage

Linked genes which are distantly located on the same chromosome and have chances of separation by crossing over, are known as incompletely linked genes and their phenomenon of inheritance is called incomplete linkage.

→ New traits occur in offsprings.

→ Zea mays – colour and shape of grain show incomplete linkage.

LINKAGE GROUPS

Linked genes → constitute linkage group

Linkage groups → corresponds to haploid number of chromosomes.

EG. Drosophila melanogaster –

4 linkage groups → 4 pairs of chromosomes.

EG. Garden pea –

7 linkage groups and 7 pairs of chromosomes.

Sex-Linkage

Transmission of X-linked and Y-linked genes from parents to offspring is called **sex-linked inheritance**.

They are of 3 types..

1. X-linked inheritance
2. Y-linked inheritance
3. XY linked inheritance

Types of Sex linkage

1. Complete sex linkage
2. Incomplete sex linkage

Complete sex linkage -

Non Homologous region

- Exhibited by genes located on **non-homologous** regions of X and Y chromosomes.
- They **inherit together** because **crossing over does not occur** in this region.
- Eg. **X linked traits** → **Hemophilia, red-green colour blindness, myopia**(near sightedness)
 - Y linked traits** → **Hypertrichosis, Ichthyosis**
(skin disorder-Dry scaly thick skin)

Incomplete sex linkage

Homologous Region

- Exhibited by genes located on **homologous regions** of X and Y chromosomes.
- **Do not inherit together** because **crossing over** occurs in this region.
- EG. X-Y linked traits
 - Colour blindness,
 - Nephritis (inflammation of nephron),
 - retinitis pigmentosa (Back wall of eye is damaged)

CROSSING OVER (MORGAN)

- Produces **new combinations** of genes by interchanging and **exchanging** or corresponding **segments** between **non-sister chromatids** of homologous chromosomes.
- Occurs during **pachytene** of **prophase I** of meiosis.
- Crossing over – steps → **Synapsis, tetrad, crossing over** and **terminalization**.
- **Universal process**– increases the chances of variation.

Morgan's Experiments showing linkage and crossing over:

Eg. Drosophila melanogaster.

- Easily be **cultured in lab**. Its life span is **short**, about **two weeks** and high rate of **reproduction**.
- Several **dihybrid cross** (like Mendel) experiments in fruit fly were done to **study genes** that are **sex linked**.
- He observed that the **two genes** did **not segregate independently** of each other and **F₂ ratio** deviated very significantly from **9:3:3:1**.

→ Morgan and his group knew that the genes were located on X chromosome and stated that when two gene in an dihybrid cross are situated on the same chromosome, then the proportion of parental combination is much higher than non-parental type.

→ Due to physical association or linkage of the two genes.

→ When genes are grouped on the same chromosome, some genes are strongly linked so they show very few recombinations. (1.3%).

→ When genes are loosely linked ie. present far away from each other on chromosome, they show more (higher) recombinations (37.2%)

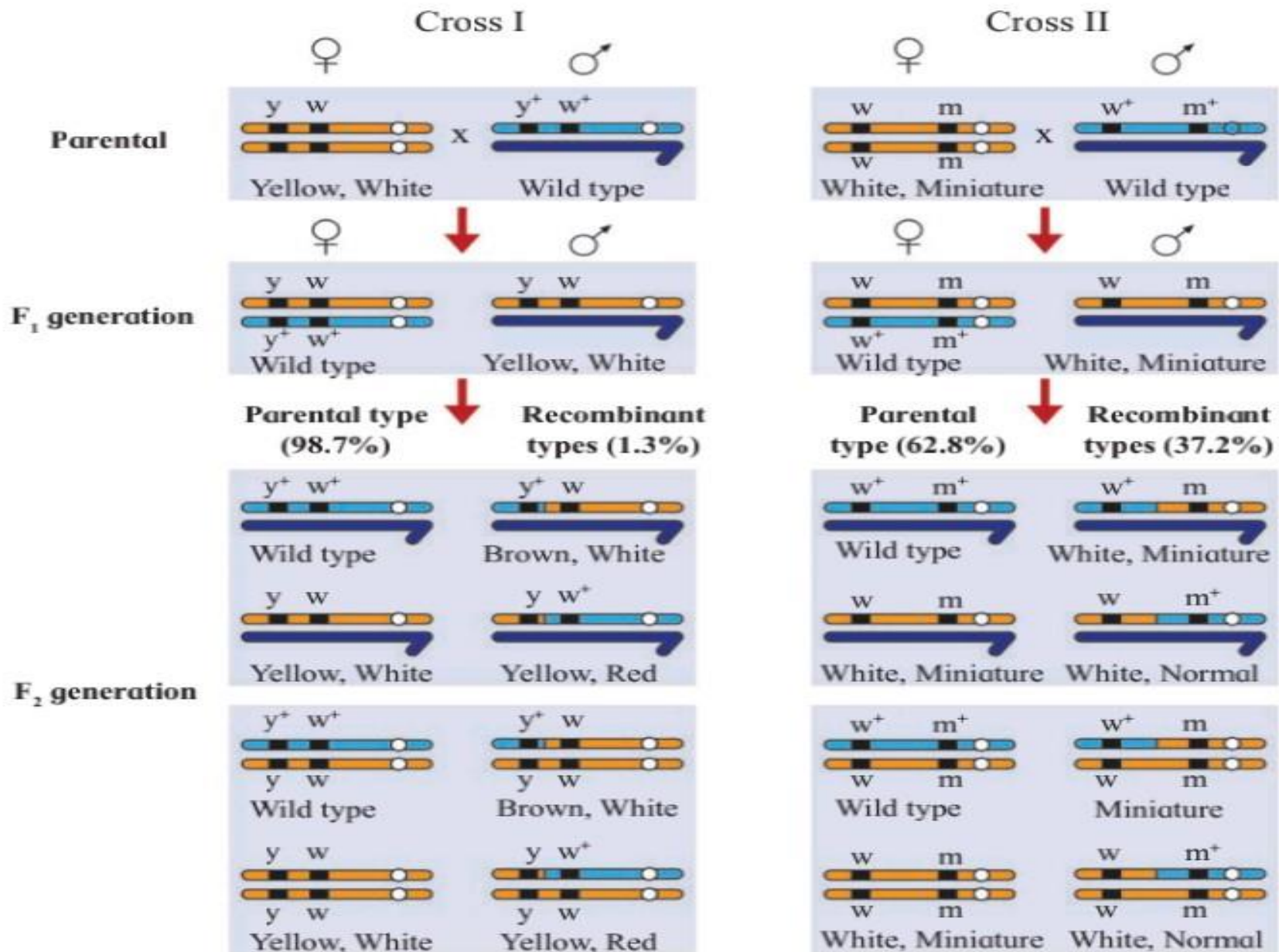


Fig : 3.8 : Linkage and crossing over

AUTOSOMAL INHERITANCE

- ❖ **Somatic** (2n) cell contains **23 pairs** of chromosomes.
- ❖ Divided functionally as **autosomes** and **sex chromosomes**.
- ❖ 1 pair - **Sex chromosomes**
- ❖ 22 pairs - **Autosomes**
- ❖ Autosomes control a **variety of traits** other than **sex**.
- ❖ **Transmission of body characters** other than the **sex linked traits** from parents to their offsprings through **autosomes**, is called **autosomal inheritance**.

AUTOSOMAL DOMINANT TRAITS:

- Widow's peak,
- Huntington's disease, etc. (Nerve cells in brain break down over the time-30's/40's)

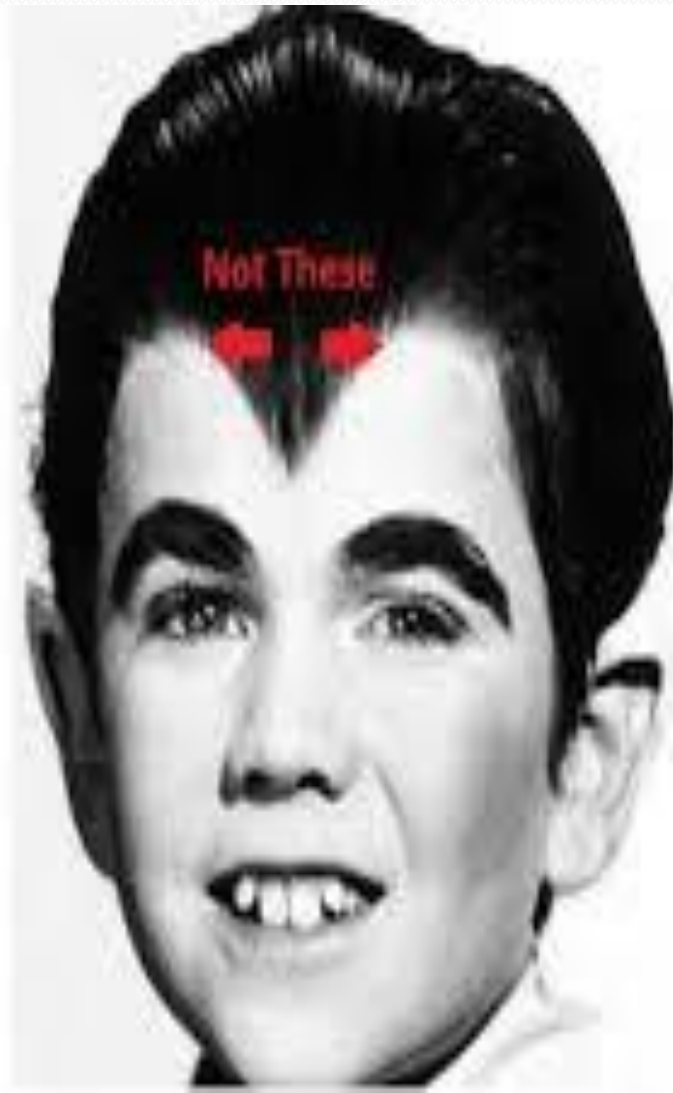
AUTOSOMAL RECESSIVE TRAITS:

- Phenyl ketonuria (PKU),
- Cystic fibrosis (affects the cells that produce mucus, sweat and digestive juices) and
- sickle cell anaemia.

A. WIDOW'S PEAK

- **V shaped** hairline on forehead.
- Determined by **autosomal dominant** gene.
- Occurs in **homozygous dominant (WW)** and also **heterozygous (Ww)** individuals.
- **Homozygous recessive (ww)** genotype have **straight hair line**. (no widow's peak)
- Both males and females have **equal chances** of inheritance.

Widow's peak



b. Phenylketonuria (PKU)

- Inborn metabolic disorder caused by recessive autosomal genes.
- Homozygous recessive genes – Phenylalanine hydroxylase enzyme is not produced.
- Enzyme is essential for conversion of ...
Phenylalanine ---> Tyrosine
- Absence of enzyme – Phenylalanine is not converted into tyrosine.
- so Phenylalanine and its derivatives are accumulated in blood and cerebrospinal fluid.(CFC)
- It affects development of brain and causes mental retardation.
- Excess is excreted in urine, hence Phenylketonuria.

SEX LINKED INHERITANCE

Definition

Inheritance of sex linked genes from parents to their offspring, is called sex linked inheritance.

There are two types of sex linked genes:

→ X-linked genes

→ Y-linked genes

X-linked gens (sex linked gene)

1. Located on **non-homologous region** of X-chromosome.
2. They have **no corresponding alleles on Y-chromosome**.
3. **Females** have **two X chromosomes**.
4. In females, **two recessive** sex linked genes are required for expression of sex linked traits.
5. If one X chromosome carries a recessive gene for sex linked trait its effect is **suppressed** by the dominant gene present on other X chromosome.
6. **Female carriers** are **physically normal**.

7. **Male** has only **one X-chromosome**.

8. If X-chromosome **carries recessive gene** for sex-linked trait, then it is **expressed phenotypically**, because there is **no dominant allele on Y-chromosome**.

9. So **X linked traits** are **more frequent in males** than females.

Examples : **Haemophilia, Colourblindness, night blindness, muscular dystrophy** etc.

Y-linked genes (Holandric gene)

1. Located on **non-homologous region** of Y-chromosome.
2. It is inherited from **male to male**.

Example : **Hypertrichosis** – Excessive development of hair on pinna of ear.

3. This character is transmitted from **father to son** directly.

COLOUR BLINDNESS

1. **X-linked recessive** disorder.
2. Person unable to distinguish between **red and green** colour as both appear grey to them.
3. Caused due to **recessive X-linked genes** (X_c) which prevents formation of **colour sensitive cells**, the **cones** in the **retina**.
4. **Dominant X linked genes** (X_C) is necessary for formation of colour sensitive cells in the retina of eye.
5. Homozygous recessive females (X_cX_c) and Hemizygous recessive male (X_cY) .Both are colourblind.

● *Inheritance of colorblindness can be studied in the following two types of marriages.*

1. → **Marriage between colour blind male with normal female**, will produce normal visioned male and female offspring in F₁.

→ The sons have normal vision but daughter will be carrier for the disease.

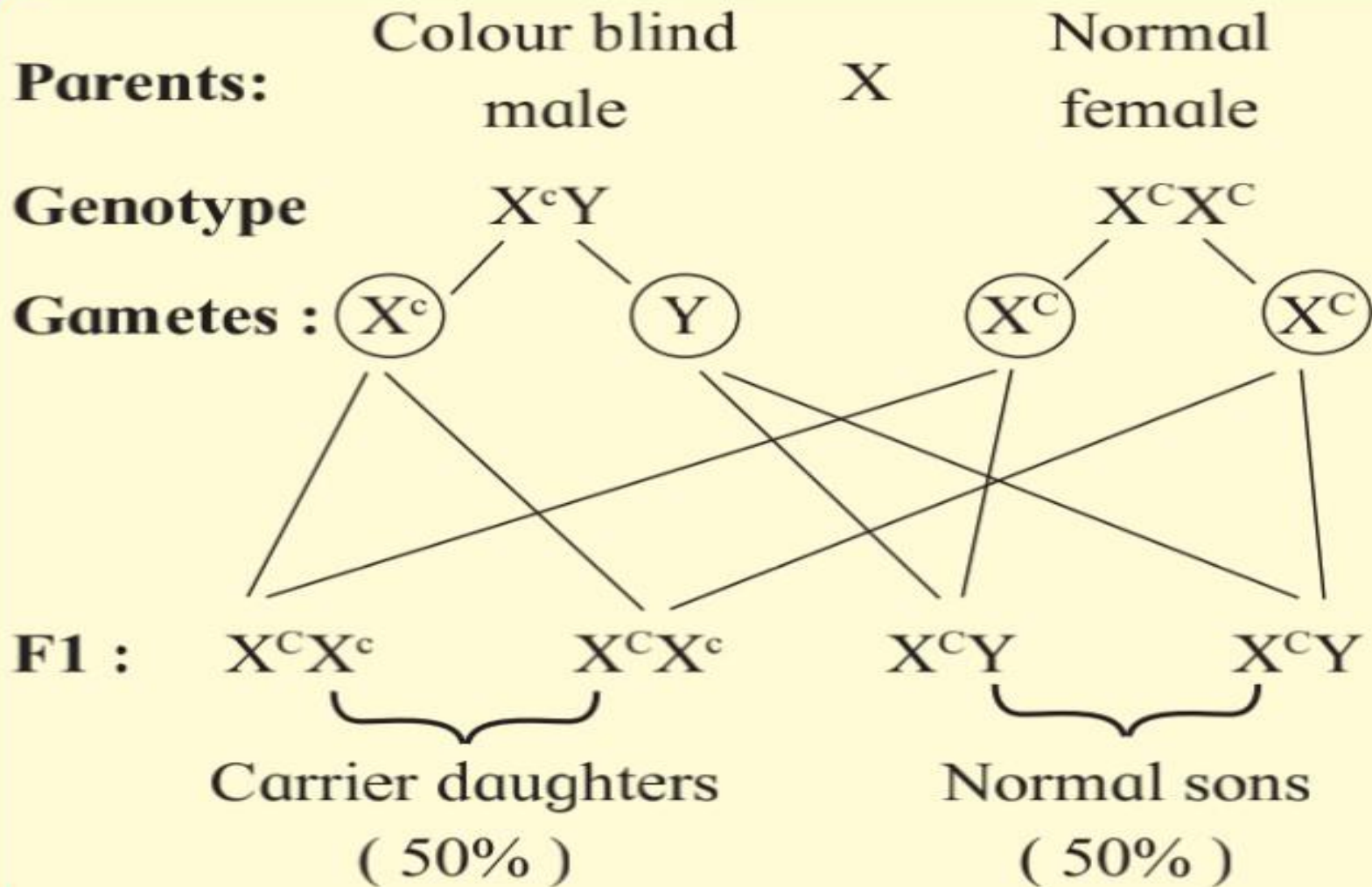


Fig. 3.10 : Sex linked inheritance (colour blindness)

Colourblindness

Eg.2 → *Marriage between carrier female (daughter) and normal male.*

- It will produce **female** offsprings with **normal vision** but **half of them** will be **carriers** for the disease.
- **Half of male** offsprings will be **normal** while **remaining half** will be **colour blind**.
- It is clear that the **X-linked recessive gene** for colour blindness is **inherited** from **colourblind father** to his **grandson** through his **daughter**.
- *This is called criss-cross inheritance.*

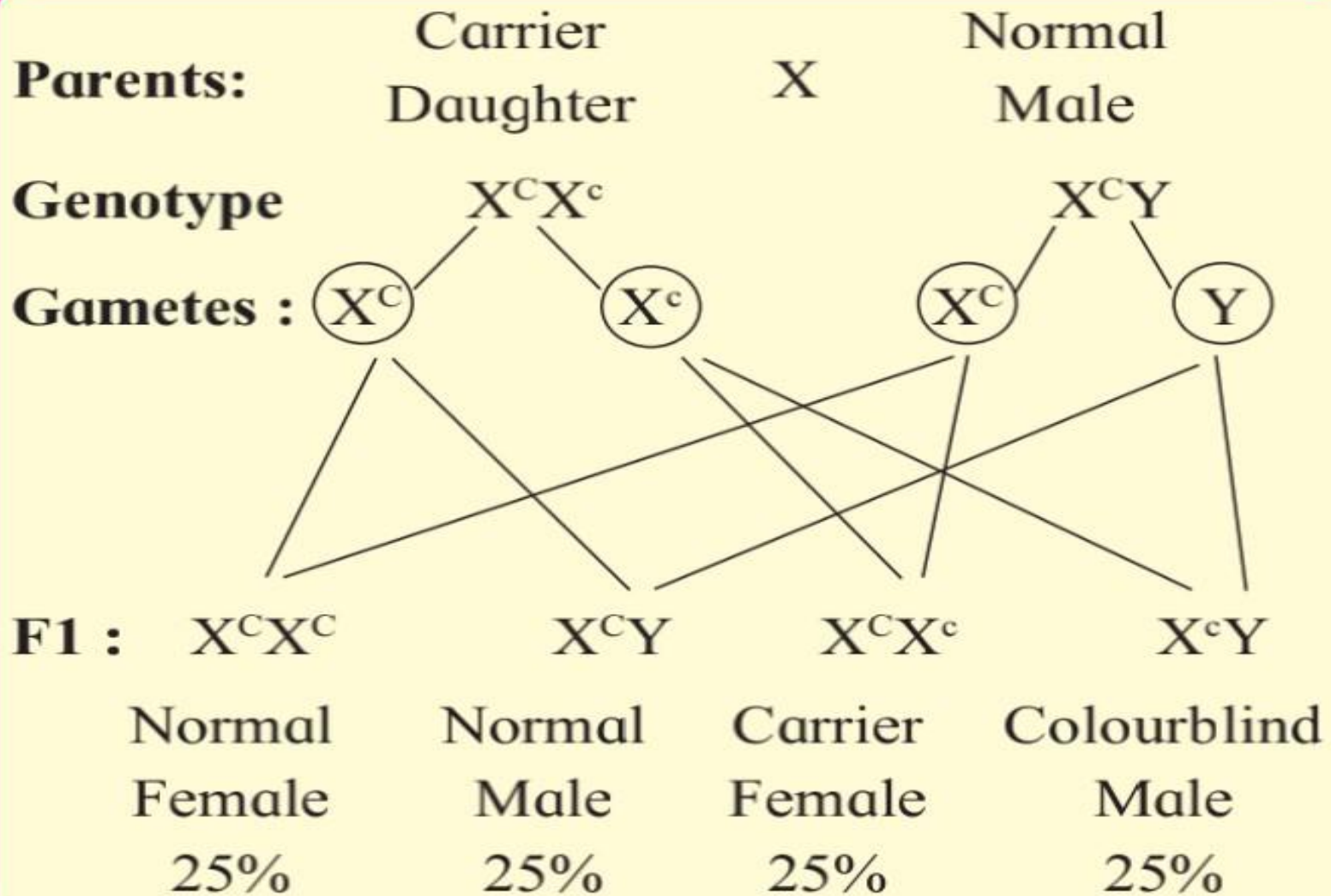


Fig. 3.11 : Sex linked inheritance (colour blindness)

HAEMOPHILIA (Bleeder's disease)

- It is **X-linked recessive disorder** in which **blood fails to clot** or coagulates very slowly.
- **Genes** for normal **clotting** are **dominant** over the **recessive** genes for haemophilia.
- The person having recessive gene for haemophilia is **deficient in clotting factors** (**VIII or IX**) in blood.
- **Minor injuries** cause **continuous bleeding** hence called **Bleeder's disease**.
- **Recessive gene** for haemophilia is located on **non homologous** region of **X chromosomes**.
- As there is **no corresponding allele** on **Y chromosome** to **suppress** its expression, **so men suffer from this disease**.

Haemophilia

1. Marriage between **Haemophilic male** and **normal female** will produce **normal male** and **female** offspring in F₁.

- The **sons** are **normal** but **daughter** will be **carrier** for the disease.

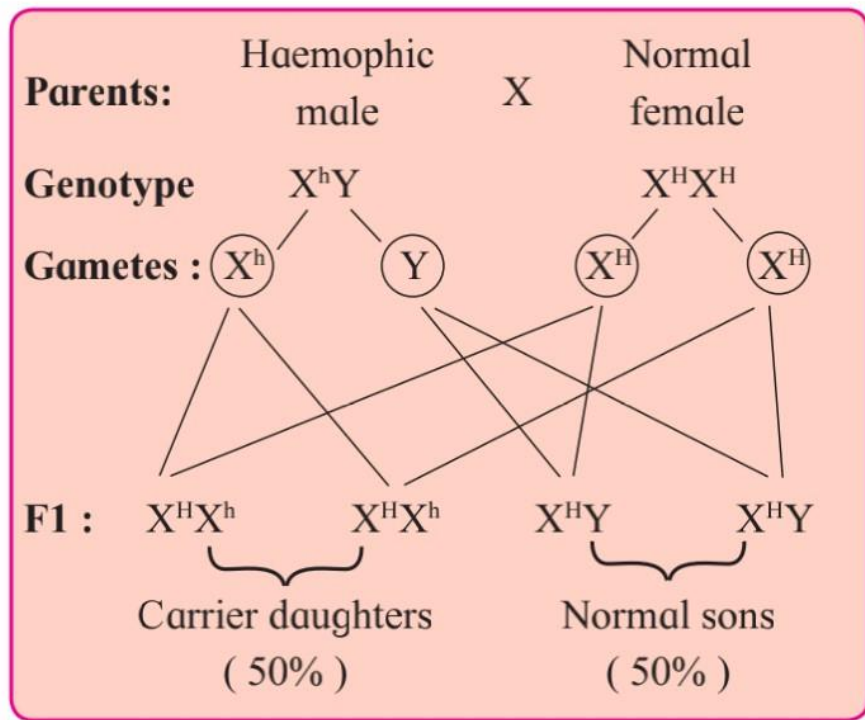


Fig. 3.12 : Sex linked inheritance (Haemophilia)

Haemophilia

Eg.2 → *Marriage between carrier female (daughter) and normal male.*

- It will produce **female offsprings** with normal blood clotting but half of them will be **carriers** for the **disease**.
- **Half of male** offspring's will be **normal** while remaining **half** will be **Haemophilic**.
- It is clear that the **X-linked recessive gene** for **Haemophilia** is **inherited** from **Haemophilic father** to his grandson through his **daughter**.
- This is called **criss-cross inheritance**.

2. Marriage between carrier female (daughter) and normal male.

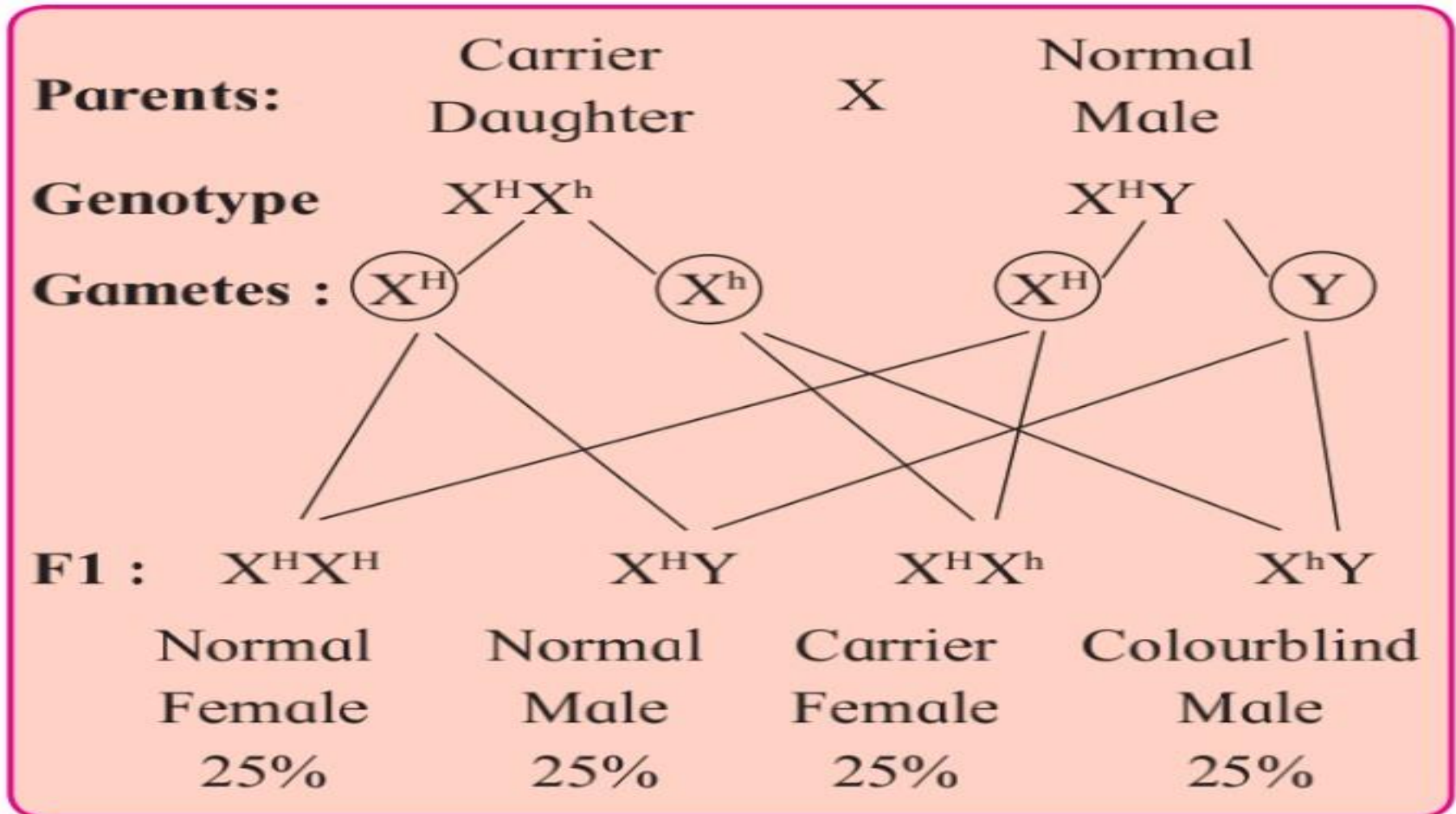


Fig. 3.13 : Sex linked inheritance (Haemophilia)

SEX DETERMINATION

- Mechanism by which sex is established is termed as **sex determination**.
- **Bisexual, hermaphrodite, monoecious** -- Some species both male and female reproductive organs are present in same organisms.
- **Monosexual, dioecious** -- Some species in which the organisms has either male or female reproductive organs.
- **Henking**-- (1891) While studying squash bug (**Anasa tristis**) found 50% of sperms receive the unpaired chromosomes while other 50% sperm do not receive it.
- He named it as **x body**.
- Later it was named as **X- Chromosome**.

SEX DETERMINATION IN HUMAN BEINGS.

Chromosomal mechanism of sex determination in human being XX-XY.

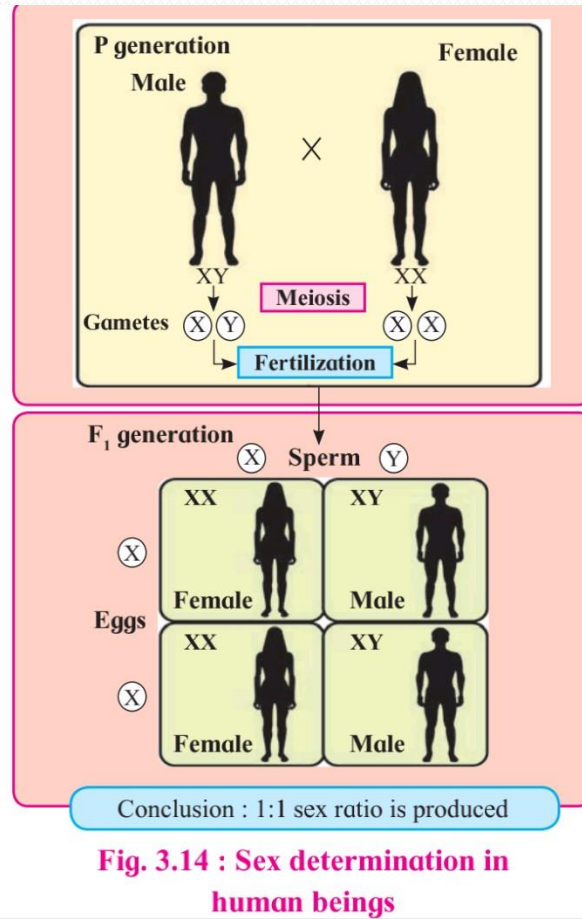
- **Nucleus of somatic cell** --46 Chromosomes or 23 pairs of Chromosomes.
- 22 pairs Autosomes and 1 pair is Sex chromosomes.
- **Female (homomorphic)** → 44 Autosomes + XX
- **Male (heteromorphic)** → 44 Autosomes + XY
- Male → testis → diploid germ cell → spermatogenesis → two types of haploid sperms →
 - 50% sperms (22 Autosomes + X chromosomes) and
 - 50% sperms (22 Autosomes + Y chromosomes).....hence heterogametic.

- Female → Ovaries → diploid germ cell → oogenesis
→ produce only one type of egg →

All eggs contain 22 Autosomes + X chromosomes.....hence homogametic.

- Sperm (X chromosome) fertilize egg (X chromosome) = Diploid zygote forms female child.
- Sperm (Y chromosome) fertilize egg (X chromosome) = Diploid zygote forms male child.
- This indicates that the sex of a child depends on the type of sperm fertilizing the egg and hence the father is responsible for determination of sex of child and not the mother.

beings



Sex determination in Birds

1. Chromosomal mechanism of sex-determination in bird is **ZW-ZZ**.
2. **Females** are **heterogametic** and produce **two types of eggs**. (**50% eggs with Z chromosomes and 50% with W chromosomes**).
3. **Males** are **homogametic** and produce **one type of sperm**.
4. **Sex of child depends on kind of egg fertilized by the sperm.**

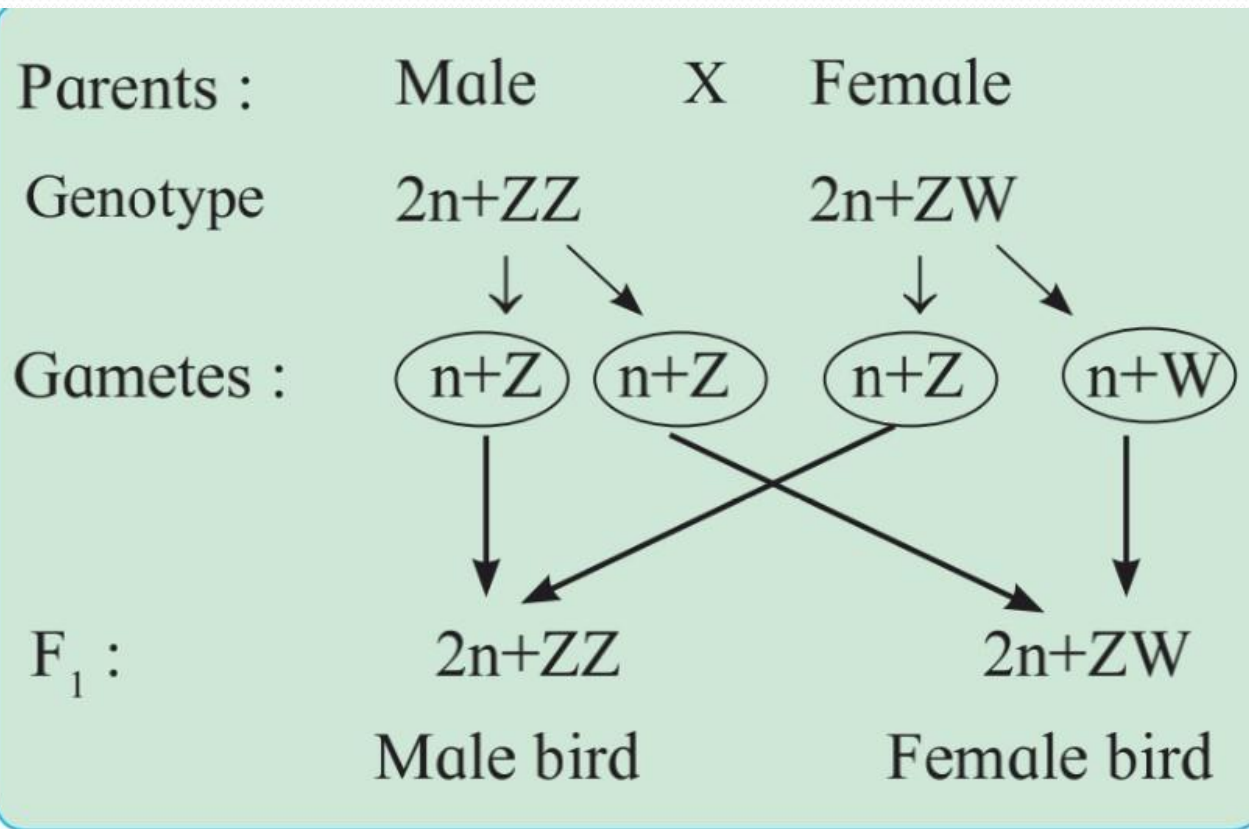


Fig. 3.15 : Sex determination in birds

Sex determination in Honey Bees

1. Chromosomal mechanism of sex determination is, **haplo-diploid** type.
2. Sex of the child is determined by the **number of set of chromosomes received.**
3. Females are **diploid** ($2n=32$)
4. Males are **haploid** ($n=16$)
5. Female produce **haploid eggs** by **meiosis.**
6. Male produce **haploid sperms** by **mitosis.**

7. If the **egg is fertilized by sperm**, the zygote develops into **diploid female**. ($2n=32$).....**Queen and Worker**.
8. **Unfertilized eggs** developed into **haploid male** ($n=16$)**Drone** by parthenogenesis.
9. Diploid female get differentiated into either worker or queen depending on the **food they consume** during their development.
10. **Larva** which get **royal jelly** as food develops into **queen**(fertile female) and other develops into **workers**(sterile females).

GENETIC DISORDERS

Mendelian disorders

Mainly caused due to **alternation** or **mutation** in the gene.

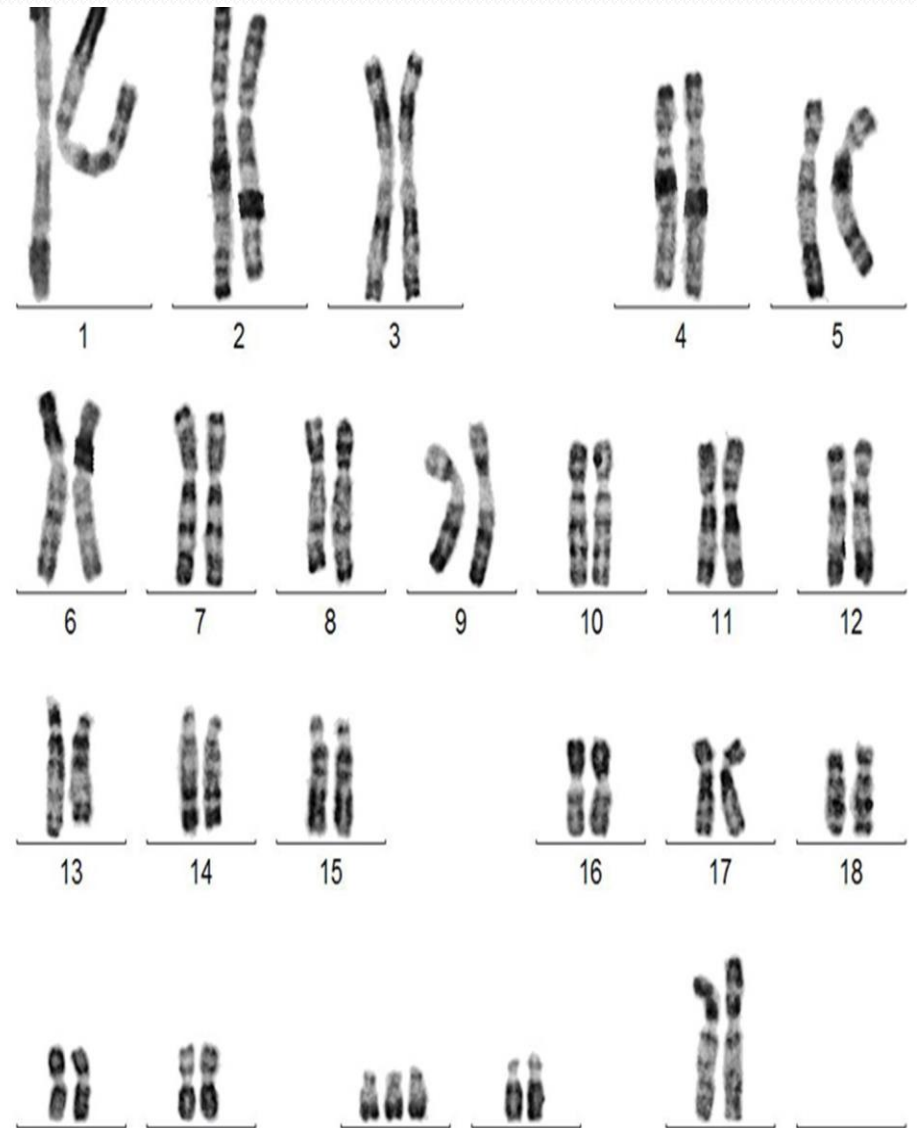
- **Thalassemia**
- **Sickle-cell anaemia**
- **colourblindness**
- **Haemophilia**
- **Phenylketonuria**

Chromosomal disorders

Mainly caused due to **absence** or **excess** of one or more chromosomes or their **abnormal arrangement**.

- **Down's syndrome**
- **Turner's syndrome**
- **Klinefelter's syndrome etc.**

Human karyotype



THALASSEMIA

- It is **autosomal, inherited recessive** disease.
- **Haemoglobin** molecule is made of **four polypeptide chains-2alpha and 2 beta chains**.
- **Synthesis of alpha chains** are controlled by two closely linked genes. (**HBA₁ and HBA₂**) **on chromosome 16**.
- **Synthesis of beta chain** is controlled by a single gene (**HBB**) **on chromosome 11**.
- Depending on chain of haemoglobin affected– classified as alpha or beta thalassemia.
- Caused due to **deletion or mutation** of gene which **codes** for **alpha** or **beta globin** chain.

→ SYMPTOMS

- Anaemia, pale yellow skin,
 - change in size and shape of RBC,
 - slow growth and development, dark urine
 - Patient needs massive blood transfusion.
- Differs from sickle cell anaemia.
- Former is qualitative problem of synthesizing few globin molecule and later is qualitative problem of synthesizing an incorrectly functional globin.

DOWN'S SYNDROME

- **Autosomal chromosomal** syndrome.
- Syndrome is caused due to the **extra copy** of **chromosome number 21st**.
- Show presence of **three copies of 21st** chromosome.
- Individual shows **47 chromosomes** instead of **46 chromosomes**.
- Occurs due to **non-disjunction** or **failure of separation** of chromosomes (autosomes) during **gamete formation**.
- Incidence is **higher in mothers** who are over **45 years old**.

Karyotype of Down's syndrome

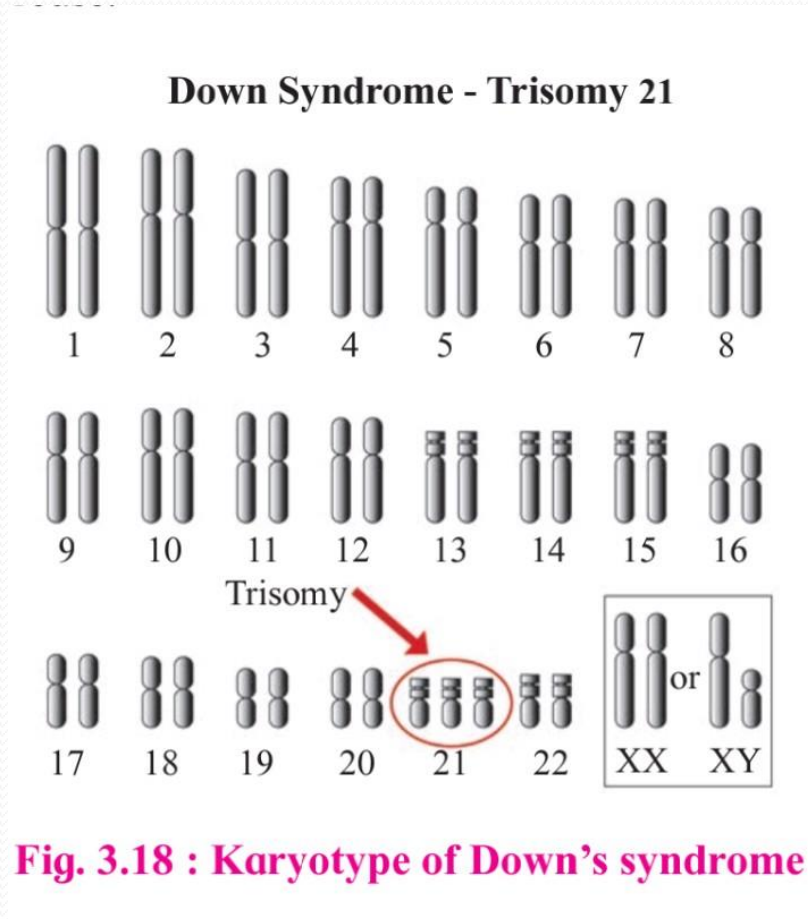


Fig. 3.18 : Karyotype of Down's syndrome

SYMPTOMS (DOWN'S SYNDROME)

1. Mild or moderate **mental retardation** and **skeletal development** is poor.
2. **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.
3. **flat hands** and **stubby fingers** and palm is broad with **single palmer crease**.
(**single line runs across the palm**)

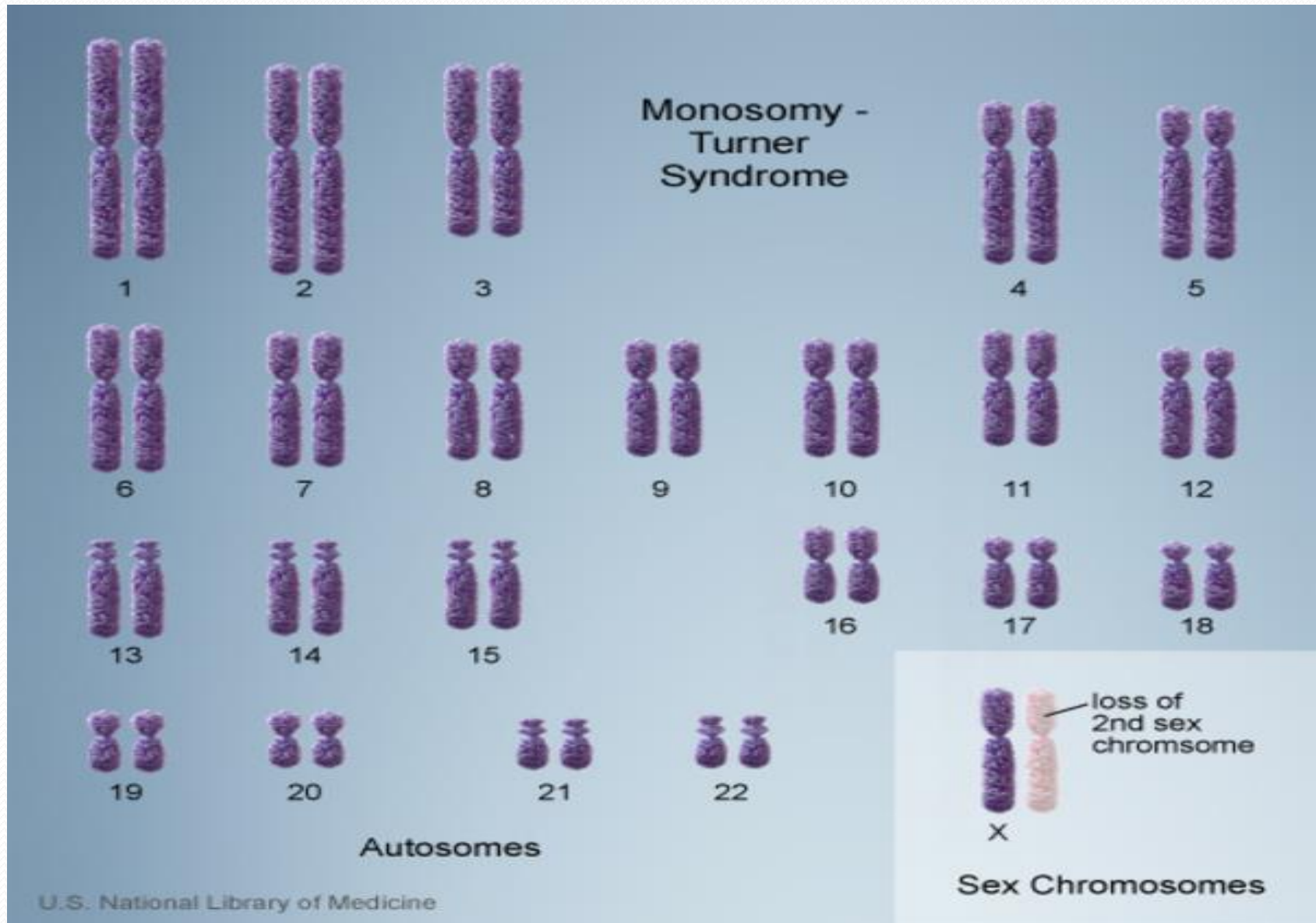
TURNER'S SYNDROME

- X monosomy / XO females.
- Sex chromosomal disorder caused due to **non-disjunction** of chromosomes during **gamete formation**.
- Individual has **44 autosomes** with **XO** sex chromosomes.
- They are **phenotypically females**.

SYMPTOMS

1. Short stature (height) and webbed neck,
2. Lower posterior hair line,
3. Broad shield-shaped chest,
4. Poorly developed ovaries and breast,
5. Low intelligence.

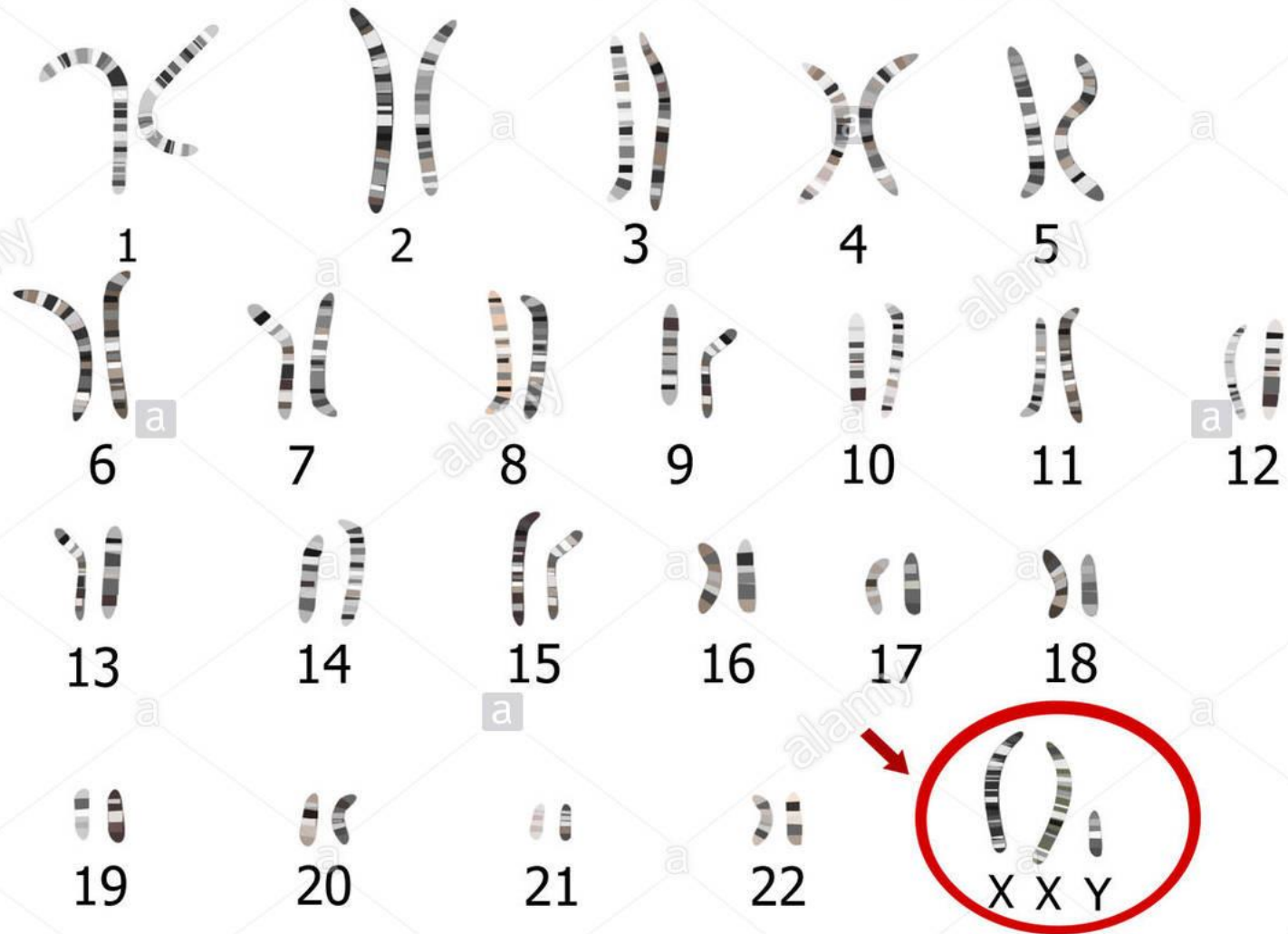
Turner Syndrome



KLINFELTER'S SYNDROME

- **XXY** males.
- Chromosomal disorder caused due to **extra X chromosome** in males.
- **Genotype** of individual is **44 + XXY**.
- They are described as **feminized males**.
- Extra chromosome is a result of **non-disjunction** of **X chromosome** during **meiosis**.

Klinefelter syndrome karyotype



SYMPTOMS

1. Individual is **male** and has over all **masculine** development.
2. Voice pitch is **harsh** and have **under developed testis**.
3. They are **tall with long arms**.
4. Feminine development (development of breast ie. **Gynaecomastia**), therefore individuals are **sterile**.

