# Biostudy with Nikhil

# Ch3) Inheritance & Variation

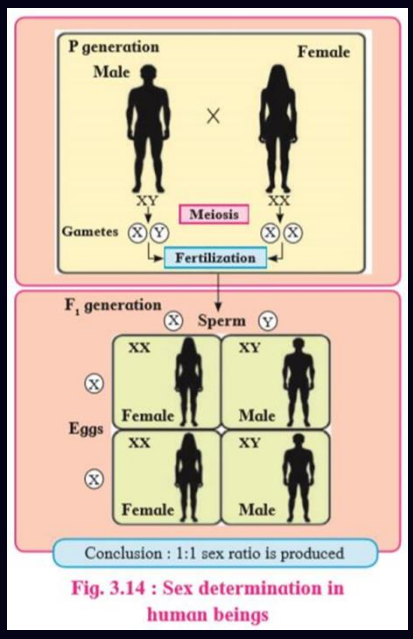


Fig. 3.14 : Sex determination in human beings

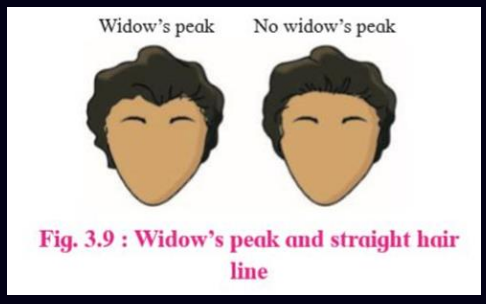


Fig. 3.9 : Widow's peak and straight hair line

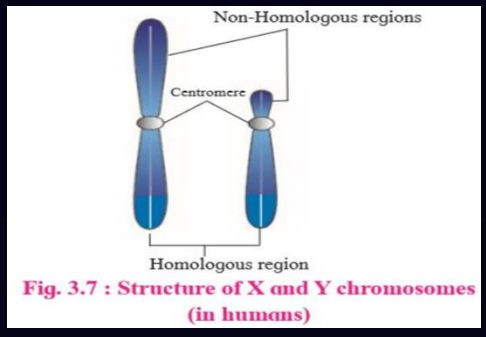
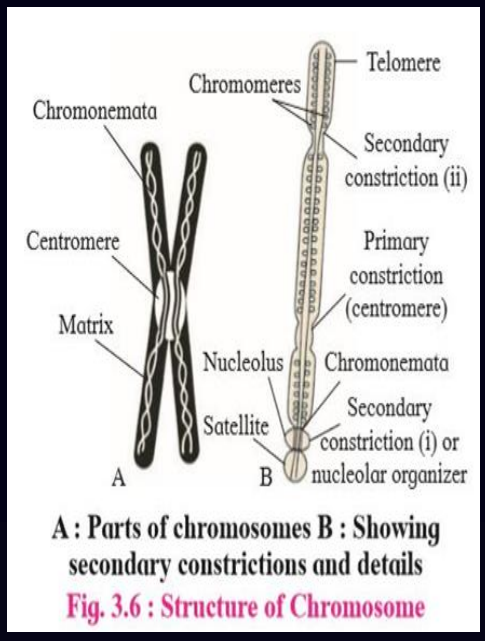


Fig. 3.7 : Structure of X and Y chromosomes (in humans)



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

Fig. 3.6 : Structure of Chromosome

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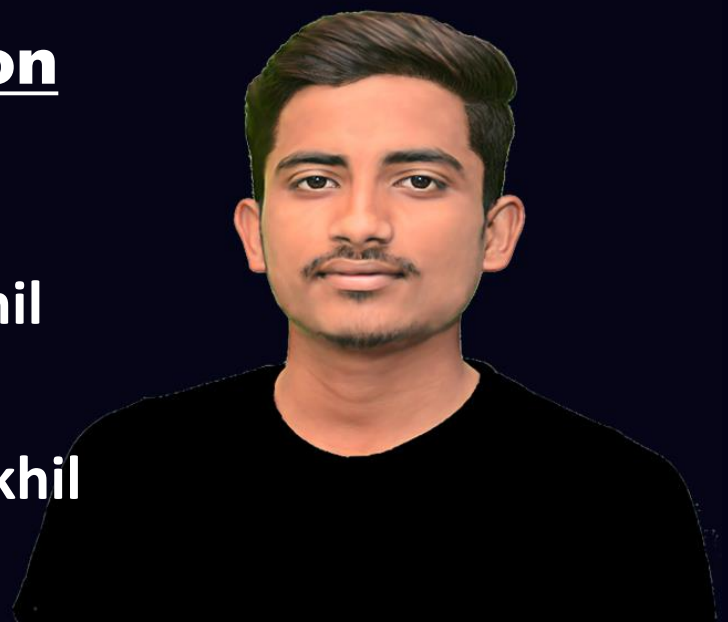


# Chp3) Inheritance and variation

## Heredity

- The process in which transfer of genetic information from one generation to next generation called.....

## Variation

- The changes / differce inbeetween parents and their offspring called.....

The branche of biology which deals with the study of Heredity and variation called genetics

- term coined William Bateson 1906
- it derived from greek word genesis which means 'to grow into'

## Father of genetics

- mendal work ⟶ mendelism
- Neomendalism ⟶ work other than mendal
- Three law
  1) law of dominace
  2) law of segrigation of gamate
  3) law of independent assortment.

## Genes

The characters are transmitted from one generation to next generation through particle called as factor → genes

## Allels

two Alternative forms of genes called as allels

## Gregor johann mendel (1822-1884)

- born on 22 July 1822
- In village in Austria poor family of farmer
- After graduation he joined the Augustinian monastery of Brunn as a teacher / priest.
- He was sent to university of Vienna
- 1856 began famous hybridization experment on gardon pea
- 1865 he present his work on paper
- Published journal "Annual proceeding of Natural history society"
- In 1866

- 1884 died

- his worked remained unnoticed
- 1900
- 1) Hugo de varies ⟶ Holland
  2) Correns (Karl) ⟶ Germany
  3) Erich Tschermak ⟶ Austria

---

| character | Dominant | Resesive |
|---|---|---|
| T.T → उच्च<br>Tt → उच्च<br>tt → पहान | The alleles Exprees in Homozygous and heterozygous state | The alleles Express in only homo-zygous state called..... |

Homozygous ⟶ similar alleles (TT)
Heterozygous ⟶ disimilar alleles. (Tt)

phenotypically ⟶ External apearance of any organism

genotypically ⟶ internal apearance of any organism

| character | dominant trait | Recesive trait |
|---|---|---|
| 1) stem height | Tall (T) | Draf (t) |
| 2) seed colour | yellow (Y) | green (y) |
| 3) seed shape | Round (R) | wrinkled (r) |
| 4) pod colour | Green (G) | yellow (g) |
| 5) pod shape | Inflated (I) | constricted (i) |
| 6) flower position | Axial (A) | Terminal (a) |
| 7) flower colour | coloured (C) | White (c) |
| 7)a) seed coat colour | coloured (C) | White (c) |

# Why mendel select garden pea for his experiment ?

Garden pea (Pisum sativum L)

- Garden pea is annual plant
- it is small herbaceous and produce many seeds
- It is naturally self pollinating Bisexual plant
- maximum contrasting characters
- No intermediate character
- easy to emasculation
- produce fertile offspring

Emasculation
⇒ Removal of stamen before Anthesis

# 1) Monohybrid cross

The cross between two homozygous individual distict/defering in between one pair of constracting character.

female → Tall (TT) × Drawf (tt) → male

T t

f1 ← Tt → gammate

| ♀ \ ♂ | T | t |
|---|---|---|
| T | TT | Tt |
| t | Tt | tt |

→ f2 generation

phenotypic ratio → 3 : 1
genotypical ratio → 1 : 2 : 1

1) Genotypic ratio:

1 : 2 : 2 : 4 : 1 : 2 : 1 : 2 : 1

1 ⟶ pure yellow Round YYRR

2 ⟶ YYRr

2 ⟶ YyRR

4 ⟶ YyRr

1 ⟶ Yyrr

2 ⟶ Yyrr

1 ⟶ yyRR

2 ⟶ yyRr

1 ⟶ green wrinkled yyrr

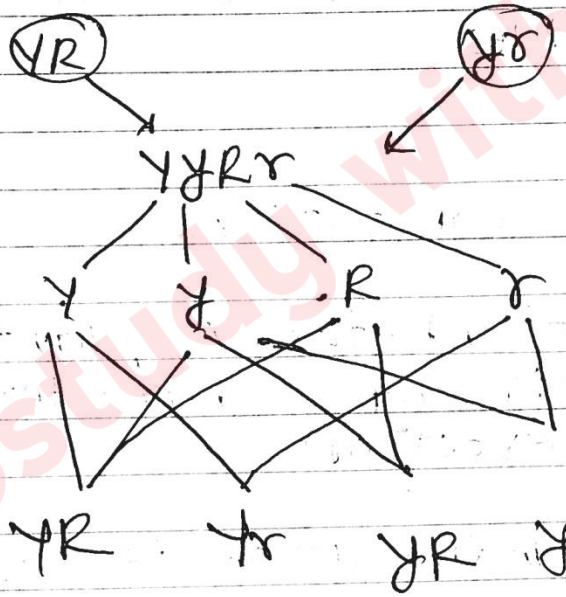
| ♀ \ ♂ | YR | Yr | yR | yr |
|---|---|---|---|---|
| YR | YYRR | YYRr | YyRR | YyRr |
| Yr | YYRr | Yyrr | YyRr | Yyrr |
| yR | YyRR | YyRr | yyRR | yyRr |
| yr | YyRr | Yyrr | yyRr | yyrr |

## 2) Dihybrid cross

The cross inbetween Homozygous individual differing (distict) in two pair of constrating character

1) seed shape (RR) (rr)
2) seed colour (YY) (yy)

YYRR × yyrr




| ♂ / ♀ | YR | Yr | yR | yr |
|---|---|---|---|---|
| YR | YYRR | YYRr | YyRR | YyRr |
| Yr | YYRr | YYrr | YyRr | Yyrr |
| yR | YyRR | YyRr | yyRR | yyRr |
| yr | YyRr | Yyrr | yyRr | yyrr |

phenotypic ratio

9:3:3:1

1) Yellow Round → 9
2) yellow wrinkled → 3
3) green Round → 3
4) green wrinkled → 1

# Mendels Laws of Inheritance

## 1) Law of dominance

- In monohybrid crosses, the phenotypic characters are controlled by discrete units, called factors.
  In a dissimilar pair of factors, one member of the pair dominates (i.e. dominant) over the other (i.e. recessive).

- The law of dominance is used to explain the expression of only one of the parental characters of a monohybrid cross in $F_1$ and the expression of both in $F_2$.

- Statement of Law of Dominance :-

  When two homozygous individuals with one or more sets of constrasting characters are crossed, the allels (characters) that appear in $F_1$ are dominant and those which do not apperar in $F_1$ are recessive.

# 2) Law of Segregation of gametes (purity of gamete)

- This law is based on the fact that the alleles do not show any blending/ mixing and both are alleles (characters) are recovered as such in the $f_2$ generation, though one of these is not seen at the $f_1$ stage.

- During formation of gametes these two alleles (factors) obviously separate or segregate, otherwise recessive type will not appear in $f_2$.

- The gametes which are formed are always pure for a particular character (unit).
A gamete may be dominant or recessive factors but not both. That's why it is called as Law of segregation of gametes.

**Statement of Law of Segregation**

"when hybrid ($f_1$) forms gametes, the alleles segregate from each other and enter in diff. gametes".
The gametes formed are pure in that they carry only one allele each (either dominant allele or recessive alleles). hence, this law is also described as Law of purity of gamates.

# 3) Law of Independent Assortment

- This law is based on dihybrid cross. It is basic principle of genetics developed by a mendal.
- It describes how diff. genes or alleles present on a separate chromosomes independently separate chromosomes independently separate from each other, during formation of gametes.
- In dihybrid cross, $F_2$ phenotypic ratio 9:3:3:1 indicates that the two pair of characters behave independent of each other.

**Statement of Low of Independent Assortment**

"when hybrid passesing two (or more) pairs of constrasting factors (alleles) forms gametes, the factors in each pair segregate independently of the other pair"

# Back cross & Test cross

## Back cross

The $F_1$ individuals obtained in a cross are usually selfed to get the $F_2$ progeny. They can also be crossed with one of the two parents from which they were derived (either recessive or dominant). Such a cross is known as back cross.

## Test cross

The cross of $F_1$ hybrid with the homozygous recessive parent is known as a test cross.

It is used to test whether an individual is homozygous (pure) or heterozygous (hybrid).

Test cross is easy, simple, repeatable and predictable.

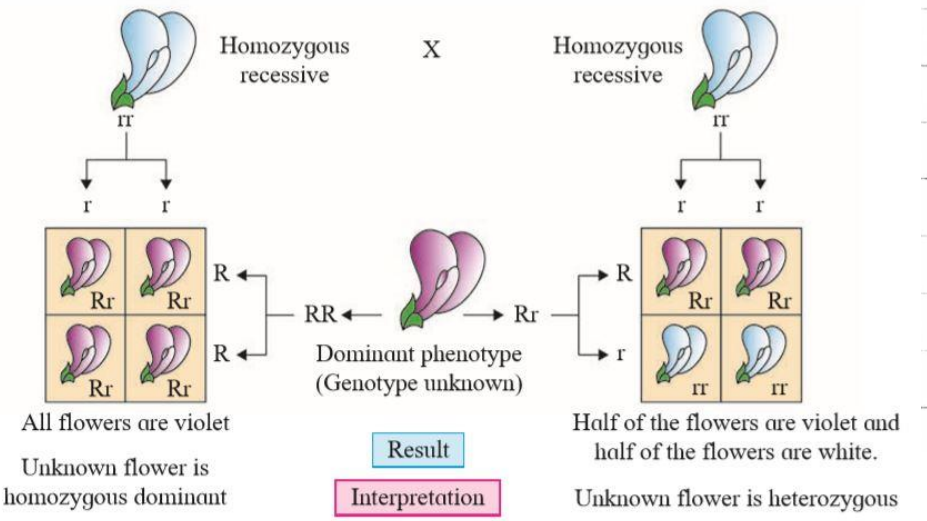



Fig. 3.1 : Graphical representation of test cross

# Deviation from mendelian ratio ( Neomendilism)

1) Gene interaction
— interaction between genes

**Intragenic**

— The interaction between alleles of an same gene
or
present on same gene called.....

1) Incomplete dominance
2) co-dominance
3) multiple alleles

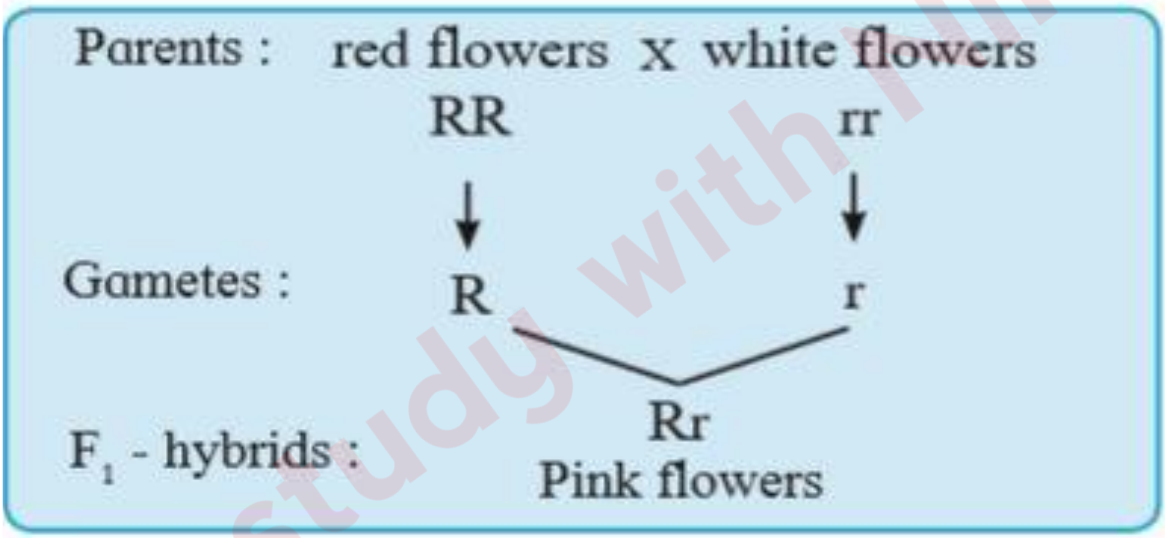
**Intergenic**

— The interaction between alleles present on two different genes called.....

1) pleotropy
polygenic

# 1) Incomplete dominance

In Incomplete dominance both the genes of an allelomorphic pair express themselves partially one gene cannot suppress the expression of other gene completely




**$F_2$ Generation : Selfing of $F_1$**

| ♀ \ ♂ | R | r |
|---|---|---|
| R | RR<br>red | Rr<br>pink |
| r | Rr<br>pink | rr<br>white |

**Result :**

Genotypic ratio - 1RR : 2Rr : 1rr

Phenotypic ratio - 1Red : 2 Pink :1 White

## 2) Co- dominance

In co-dominace both genes ~~are~~ of an allelomorphic pair express themselves equally in F1 hybrids

They express themselves independently even if present together

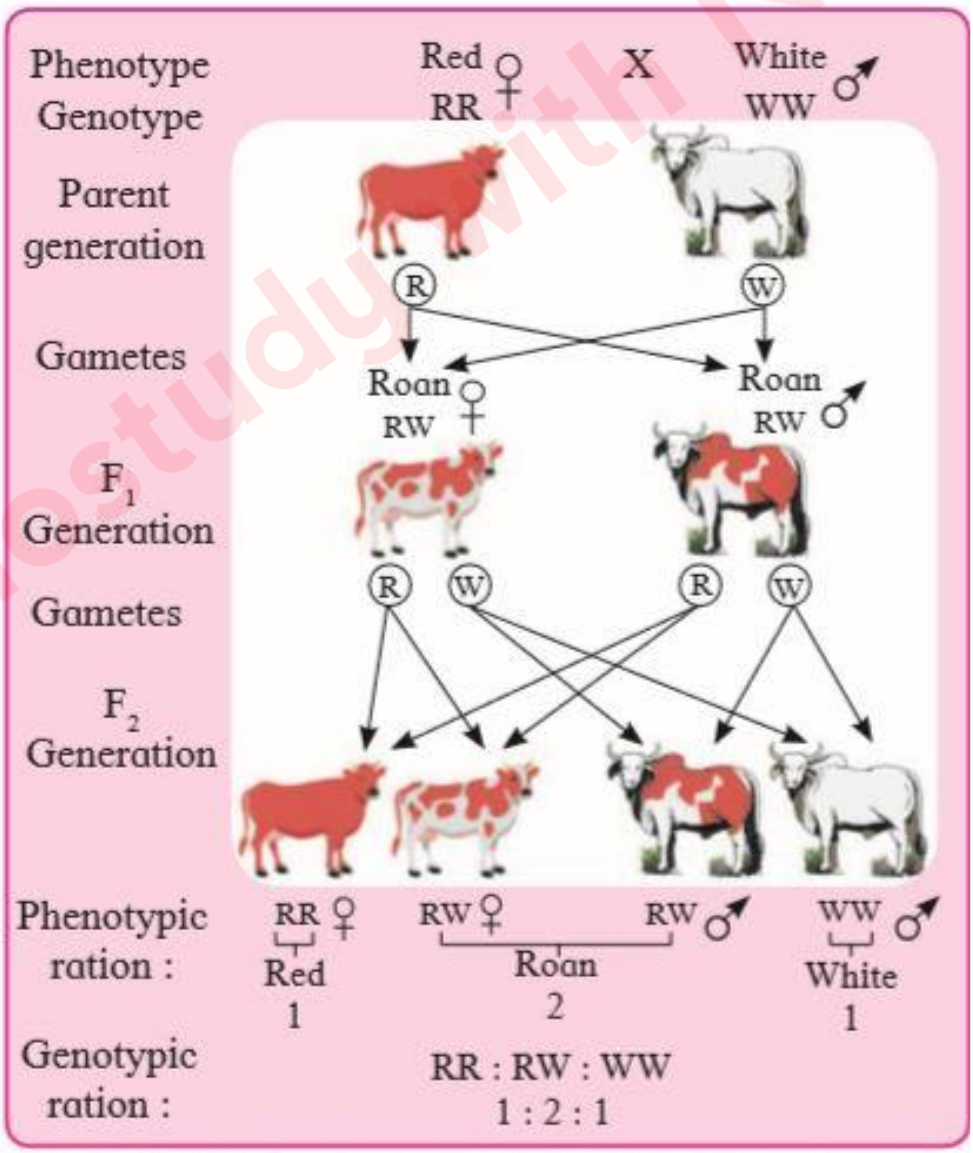



**Fig. 3.2 : Representation of co-dominance in cattle**

# 3) Multiple Allels

- More than two alternative froms of gene in a population occupying same locus of chromosome called.....

eg

**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 |

2) Blood group

| phenotype | genotype |
|---|---|
| A | $I^A I^A$, $I^A I$ |
| B | $I^B I^B$, $I^B I$ |
| AB | $I^A I^B$ |
| O | $I I$ |

A, B, AB, O ⟶ ABO

1) multiple allels do not undergoes over,

2) multiple allels aries by mutation of wild type gene

# 1) Pleiotropy

- When single gene control two different trait called.....

eg ⇒ sickle cell anemia

Normal $Hb^A Hb^A$ carrier $Hb^A Hb^S$
infected $Hb^S Hb^S$

$Hb^A Hb^S$ × $Hb^A Hb^S$

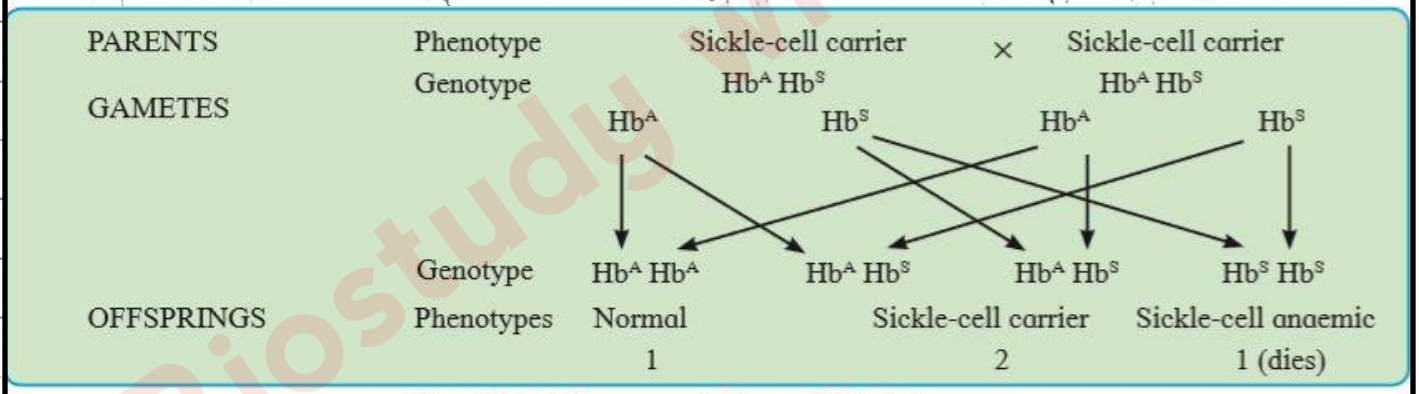



**Fig. 3.4 : Representation of Pleiotropy**

# The Chromosomal Theory

→ "chromosomal theory of inheritance" was discovered by Sutton and Boxeri in 1904.

- It is the theory of genetics which identifies chromosomes as the carrier of genetic material.

① Gametes (sperm and egg) carry all the hereditary characters. They are link between parents and offspring.

② Nucleus of gametes contains chromosomes which carry all the heriditary characters.

③ chromosomes are found in pairs in somatic or diploid cells.

④ During gamete formation homologous chromosomes pair and segregate or separate indepently at meiosis. Thus each gamete contains only one chromosome of a pair.

⑤ During fertilization, the union of sperm and egg restore the diploid number and of chromosome.

## ② CHROMOSOMES (chroma - colour, soma - body)

→

* chromosomes are filamentous bodies present in the nucleus
- They are visible only cell division.
- chromosome have the ability of self replication and they play an important role in heredity, variation, mutation & evolution

- chromosomes which are morphologically and genetically identical are called homologous chromosomes.

## Number

- All the individual of species usually have a some nu. of chroso.
- presence of whole sets of chromosomes is called euploidy.
- It includes (n) haploid (one set of chromosomes), (2n) diploid (two set of chromosomes), triploid (3n), tetroploid (4n) etc.

for example) In human beings each somatic (body cell) cell contains two sets of chromosomes (diploid 2n=46) and gametes contain one set of chromosomes (haploid n=23).

out of 23 pair of chromosomes 22 pair are autosomes and one pair is sex chromosomes.

## Size

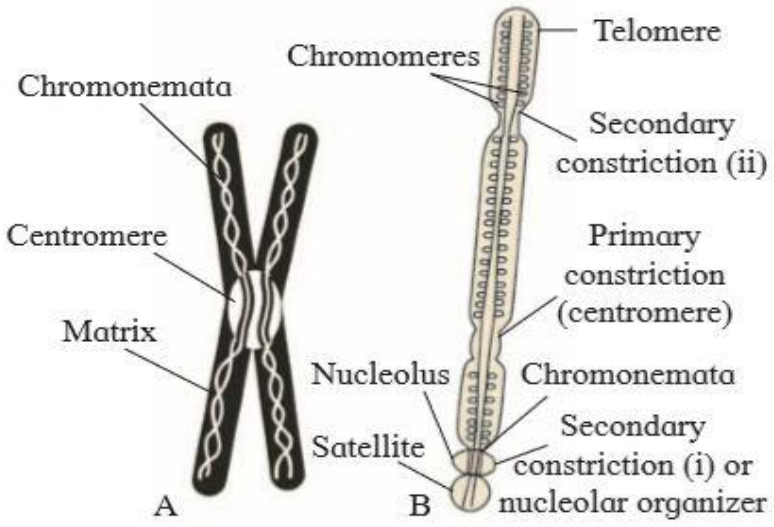
- The size of chromosomes varies from species to species. each metaphase chromosome varies from 0.1 to 33um in length and 0.2 to 2um in thickness.

## Shape

- The shape varies according to the stages of cell division In interphase, chromosomes are in the form of chromatin network while in metaphase they show maximum condensation and appear short and thick....

# Structure

- A metaphasic chromosomes has two identical halves called sister chromatids.
- Each chromatids are made up of sub-chromatids called chromon ma
- The cromatids lie side by side and are held together at one poin called the centomere.
- the centromere is also called as primary constriction.
- During the cell division the spindle fibres are attached at the centromere, Besides the primary constriction, additiona narrow areas called secondary constriction.
- The part of the chromosome beyond the nucleolar organi is short, spherical and is called satellite.
- It has the unique property in that it prevents the ends of the chromosomes from sticking together but attaches to the nuclear envelope.
- the surface of a chromosome bears number of small swellings called chromomeres...




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

**Fig. 3.6 : Structure of Chromosome**

# TYPES OF CHROMOSOMES

## 1) Metacentric

1) If the chromo centro mere is situated in the middle of the chromosome it is called as metacentric chromosome

2) The two arms of the chromosome are nearly equal.

3) It appear 'V' shaped during anaphase.

metacentric

## 2) sub-metacentric

1) If the centromere is situated some distance away from the middle it is called sub-metacentric chromosome.

2) one arm of the chromosome is shorter than the other

3) It appears 'L' shaped in anaphase.

sub-metacentric

## 3) Acrocentric

1) If the centromere is situated near the end of the chromo.. then it is klas...

2) one arm of chromo. is very short while other is long.

3) It appear 'J' shaped in anaphase.

Acrocentric

## 4) Telocentric

1) If the centromere is situated at the tip of the chromosome the it is klas....

2) It show only one arm and appear rod shaped.

Telocentric.

# Structure of X &Y Chromosome

- X chromosome is longer than Y chromosome.
- X chromosome contains large amount of euchromatin & small amount of heterochromatin.
- The euchromatin contains large amount of DNA or genes.
- Y chromosome contains small amount of euchromatin & large amount of heterochromatin. It has less DNA hence it is genetically less active.
- X and Y chromosomes show homologous & non-homologous region homologous regions show similar genes while non-homologous show dissimilar genes.
- Non-homologous region of X-chromosome contain more genes than that of non-homologous region of Y chromosome.
- Non-homologous region of X-chromosome contain X-linked genes while non-homologous region of Y chromosome contain Y-linked genes.

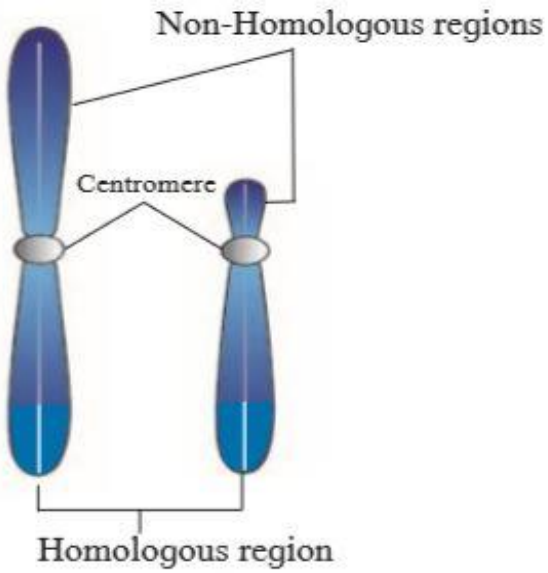



**Fig. 3.7 : Structure of X and Y chromosomes (in humans)**

# Morgan's Experiment showing Linkage & Recombination

- morgan used Drosophila melanogaster (fruit fly) for his experiments because, Drosophila can easily be cultured in laboratory.

- Its life span is short, about two weeks. more over it has rate of reproduction.

- Morgan carried out several dihybrid cross experiments in fruit fly to study genes that are sex-linked.

- The croses were similar to dihybrid crosses, as carried out by mendal in pea.

  for ex. Morgan and his group crossed yellow-bodied white eyed female to the wild type with brown bodied ~~female to the~~, red eyed males and intercrossed their $f_1$ progeny.

- He observed that the two genes did not segregate independently of each other and $f_2$ ratio deviated very significantly from 9:3:3:1 ratio.

# Linked

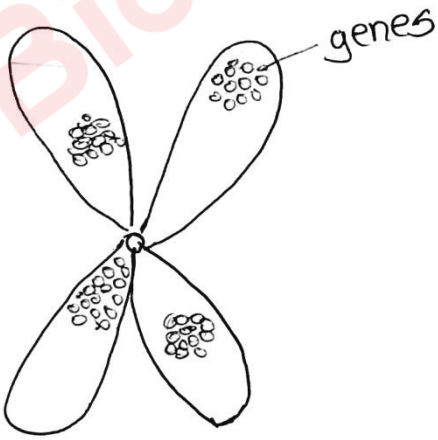
Genes present on the chromosome they have a tendency to inherit together is called linkage ""

eg ⇒ Number of linkage group in human being are 23

Linkage group can be broken by crossing over

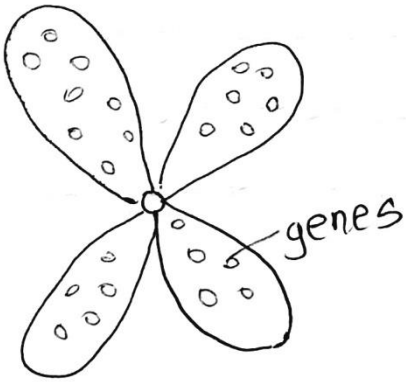
## Complete Linkage

The linked genes which are closely located in the chromosome, do not separate by crossing over and inherit together are called as complete linkage




## Incomplete Linkage

The linked genes which are widely located on the chromosome and may separate by crossing over are called as Incomple linkage



# Sex-Linked

The transmission (inheritance) of X-linked and Y-linked gene from parents to offspring called as sex-linkage. Sex-linked inheritance is of three type viz, X-linked, Y-linked and XY-linked.

## Complete Sex Linkage

It is 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.

Example of X-linked trait are - haemophilia, red-green, colour blindness, myopia (near sightedness) and for Y-linked are hypertrichosis, Ichthyosis etc.

## Incomplete Sex Linkage

It is exhibited by genes located on homologous regions of X and Y chromosomes. They do not inherit together because crossing over occurs in this region.

Example of X-Y linked trait are total colour blindness, nephritis, retinitis pigmentosa etc

# Sex-Linked Inheritance

Genes located on non-homologous region of sex chromosome but not involved in sex determination are called sex-linked genes

## X-Linked Inheritance

- Genes located on non-Homologous region of x-chromo some only are called x-linked or sex linked genes
- In humon beings are responsible for sex-linked character

eg
- Haemophilia
  Colour blindness
  nigh blindness
  myopia
  muscular dystrophy

## Y-Linked Inheritance

- Genes located on non-Homologous region of y-chromo some only are called y-linked or holandric genes

eg
- Hypertrichosis
  (excessive growth of hair on pinna of ear)

# Haemophilia

- Haemophilia is a hereditary blood disease in which blood fails to clot or clots very slowly
- haemophilia has deficiency of clotting factors (factor VIII or IX)
- It is also called bleeder's disease

| Sex | Normal | Haemophilic | Carrier |
|---|---|---|---|
| Male | $X^HY$ | $X^hY$ | —— |
| Female | $X^HX^H$ | $X^hX^h$ | $X^HX^h$ |

- If a haemophilic male marries a female with normal clotting of blood then all offspring have..... normal clotting blood but the daughter will be carriers for the disease

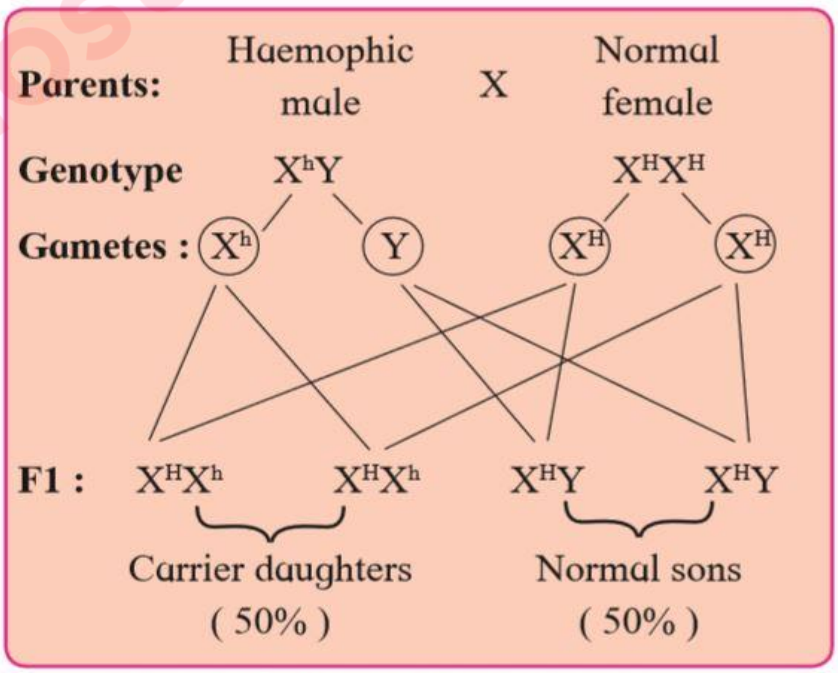



**Fig. 3.12 : Sex linked inheritance (Haemophilia)**

# 1) Colour Blindness

- In which the person cannot distinguish between red and green colour as both these colour appear grey

| Sex | Normal | Colourblind | Carrier |
|---|---|---|---|
| Male | $X^CY$ | $X^cY$ | —— |
| Female | $X^CX^C$ | $X^cX^c$ | $X^CX^c$ |

) If a blind male marries a female with normal vision then all the offspring will have..... normal vision.

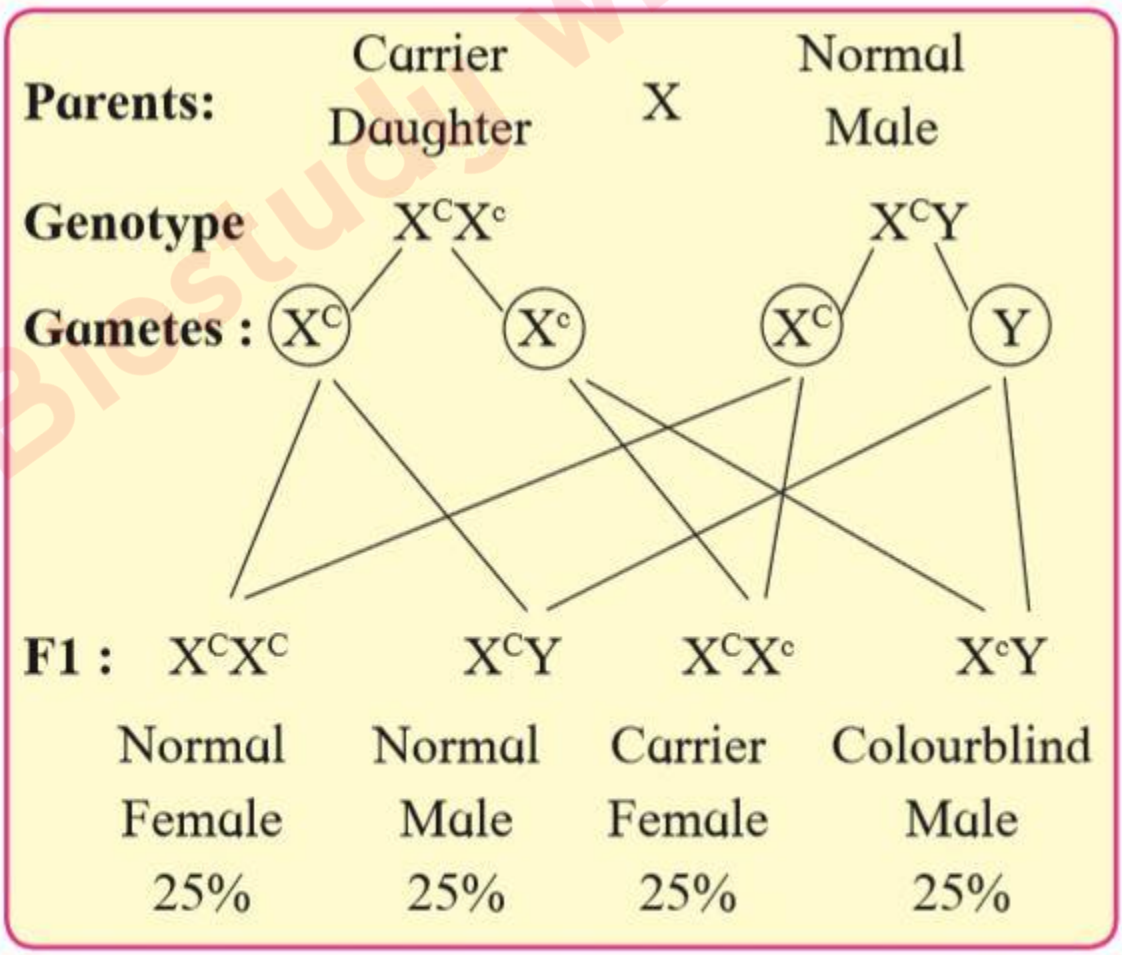



**Fig. 3.11 : Sex linked inheritance (colour blindness)**

# Sex Determination

## In Human beings

⇒

1) The chromosomal mechanism of sex-determination is of XX-YY type.

2) The human nucleus of each cell contain 46 chromosomes or 23 pairs of chromosomes. Of these 23 pair are autosomes (responsible for determination of body characters) and 1 pair is of sex chromosomes. (responsible for determination of sex.)

3)- In female two homomorphic sex chromosomes are XX & in male two heteromorphic sex chromosomes are XY.

Thus the genotype of female and male is

female : 46 chromosomes = 44 Autosomes + XX, (sex chromosomes)

Male : 46 chromosomes = 44 Autosomes + XY (sex chromosomes)

## Mechanism of sex determination

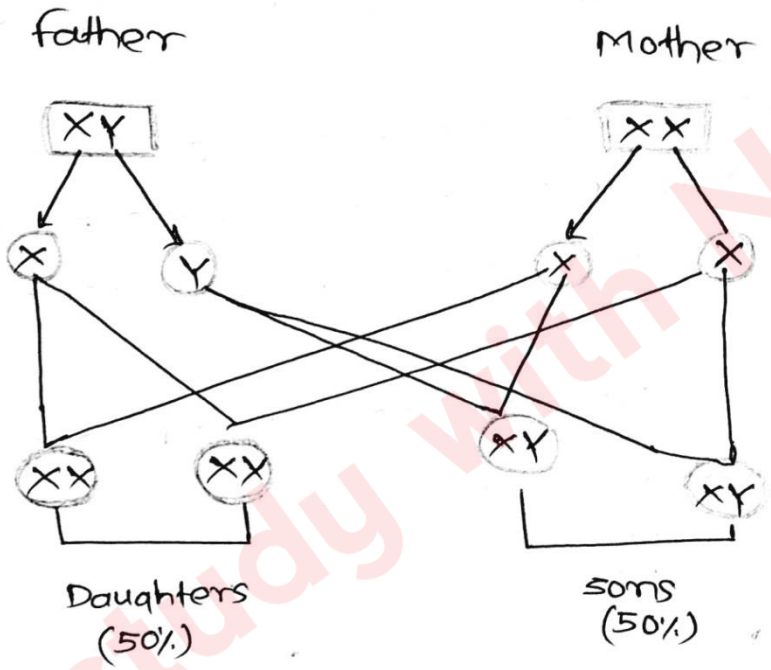
1) During gamete formation, the diploid germ cells in the testes and ovaries undergo meiosis to produce haploid gametes (sperm & eggs).

2) The human male is heterogametic and produce to types of sperm - One type of sperm contain 22 autosomes and one X-chromosomes and the other type of sperm contain 22 autosomes and one Y-chromosome.

3) The human female is homogametic and produce only one type of eggs containing 22 autosomes and one X-chromosomes.

During fertilization if X- containing sperm fertilizes the egg having X-chromosome then resulting offspring with XX chromosome is a female.

) If Y-containg sperm fertilizes the egg having X-chromosome then resulting offspring with XY chromosome is a male.




6) This indicates that the sex of the child depends on the types of sperm fertilizing the egg and thus the father is responsible for sex of the child and not the mother.

7) chromosomal mechanism of sex determination is called heterogamesis. It may be male heterogamety or female is heterogamety.

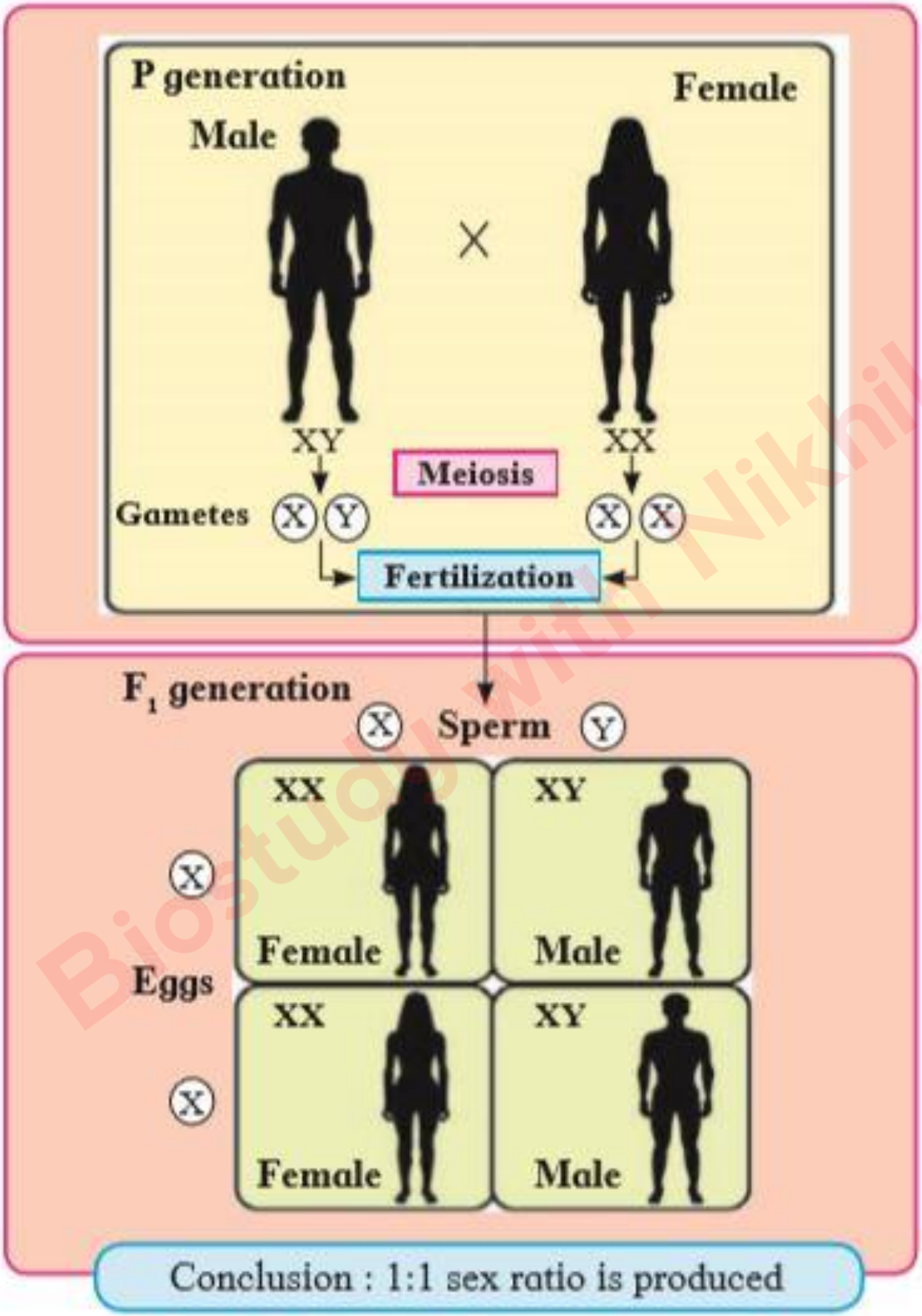



**Fig. 3.14 : Sex determination in human beings**

# Sex determination in honey bees

1) sex is determined by the number of sets of chromosomes received by an individual. such type of sex determinatn is called halpo-diploid sex determination system.

2) the fertilized egg develops as a female offspring (may be queen & worker). It shows diploid (2n=32) no. of chromosomes.

3) An unfertilized egg develops as a male (drone) by means of parthenogenesis., thus the dromes have haploid (n=16) no. of chromosomes. the dromes produce sperm by mitosis.

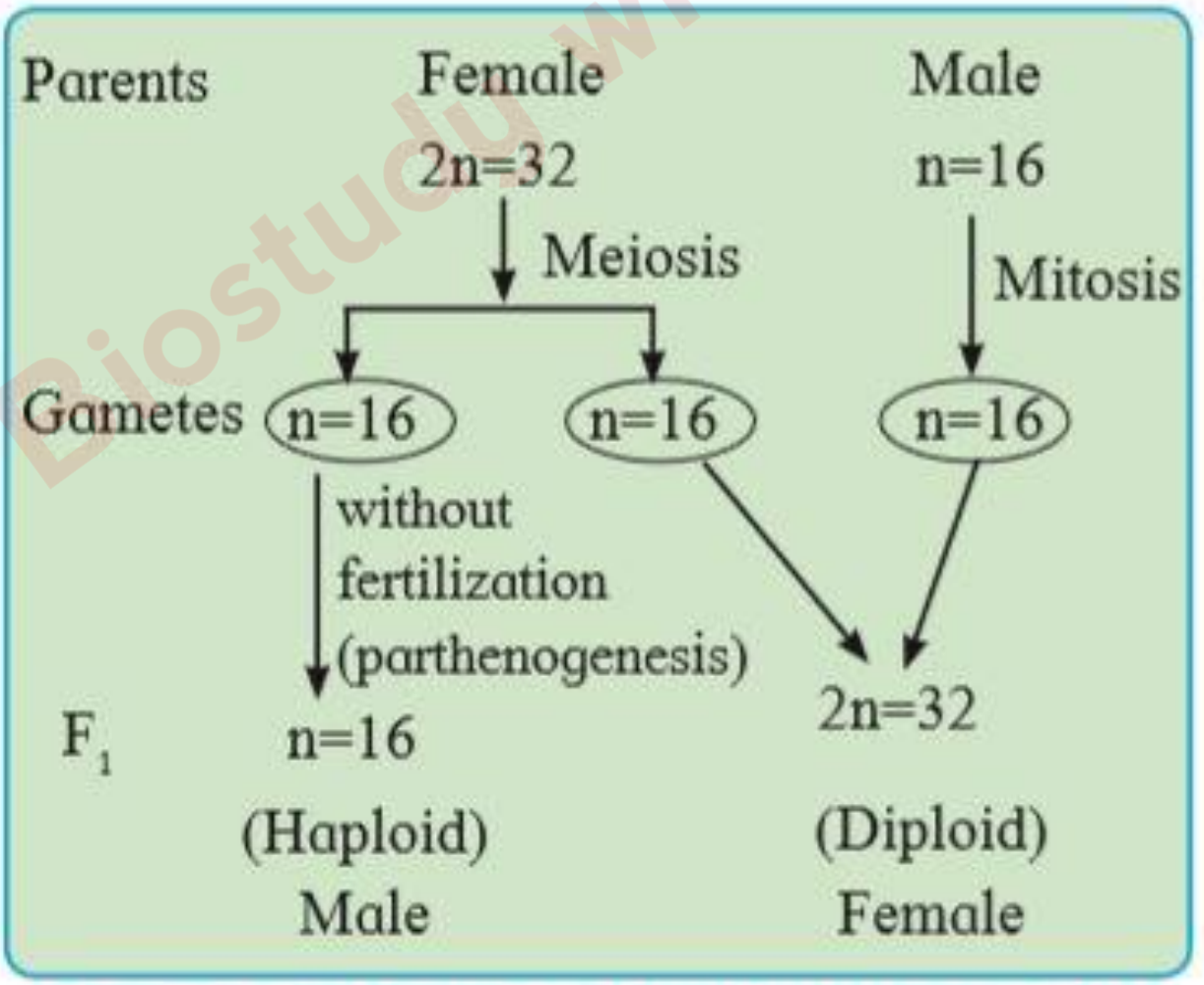



Fig. 3.16 : Sex determination in honey bee

# Sex determination in birds

4) In this type males are homogametic and have ZZ sex chromosomes and female are heterogametic with ZW pair of sex chromosome.

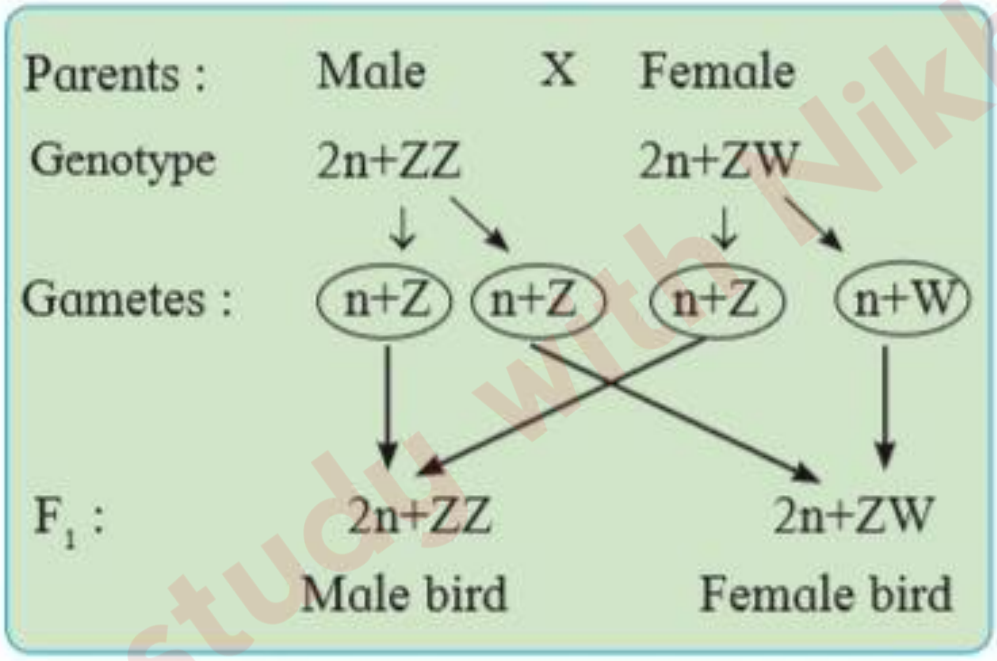



**Fig. 3.15 : Sex determination in birds**

# Autosomal Inheritance

Human somatic (2n) cell contains 23 pairs of chromosome they can be, devide functionally as autosomes and sex chromosome. A single pair of chromosome is involved in sex determination and remaining 22 pairs are called autosomes. Autosomes control a variety of traits other than sex. These traits are called autosome linked traits.

Transmission of body characters other than the sex linked traits from parents to their offsprings through autosomes, is called autosomal inheritance.

Some characters are influenced by dominant genes while some other are by recessive genes, present on autosomes. for examples,

- Autosomal dominant traits like widows peak and Huntington's disease etc.
- Autosomal recessive traits like phenyl ketonuria (PKU) cystic fibrosis and sickle cell anaemia.

## A) Widows Peak

A prominent 'V' shaped hairline on forehead is described as widows peak. It is determined by autosomal dominant gene. widow's peak occurs in homozygous dominant (WW) and also heterozygous (Ww) individuals. Individuals with homozygous recessive (ww) genotype have a straight hair line (no widows' peak). Both males and females have equal chance of inheritance.


**Fig. 3.9 : Widow's peak and straight hair line**

## B) Phenylketonuria

It is an inborn metabolic disorder caused due to recessive autosomal genes. When recessive genes are present in homozygous condition, phenylalanine hydroxylase enzyme is not produced. This enzyme is essential for conversion of amino acid phenylalanine into tyrosine. Due to absence of this enzyme, phenylalanine is not converted into tyrosine. Hence, phenylalanine and its derivatives are accumulated in blood & cerebrospinal fluid (CSF). It affects development of brain and causes mental retardation.

# Genetic Disorders

genetic disorders are broadly grouped into two categories as, Mendelian disorders and cromosomal disorders.

## Mendelian Disorders

caused by

due to alteration or mutation in the gene

ex. colourblindness, thalassemia, haemophilia, phenylketonuria, etc.

## Chromosomal Disorders

caused by

due to absence or excess of one or more chromosomes or their abnormal arrangments.

ex. Down's syndrome, Turner's syndrome, Klinefelter's syndrome etc.

### Thalassemia

- Thalassemia is an autosomal, inherited disease
- Haemoglobin molecule is made of four polypeptide chain
  - 2 alpha ($\alpha$)
  - 2 beta ($\beta$) chain

- synthesis of alpha chain are controlled by two closely linked genes (HBA1 & HBA2) on chromose 16
- synthesis of beta chain is controlled by a single gene (HBB) on chromome 11

- Depending upon which chain of haemoglobin is affecte thalassemia is classified as alpha-thalassemia & beta-Thalassemia

# Chromosomal disorders

## 1) Down's syndrome (21th th Trisomy)

- John Langdon Down who first described this autosomal chromosomal disorder in 1866
- caused due to extra copy of chromosome number 21th

It shows presence of three copies of 21th chromosome instead of homologous pair

- These individuals have 47 chromosome

21th trisomy occurs due to non-disjuction of chromosome during the ete formation.

## 2) Klinefelter's syndrome (XXY males)

- It is chromosomal disorder caused due to extra X chromosome in males
- This genotype of individual is 44 + XXY
- Extra chromosome is a result of non-disjunction of x-chromosome during meiosis
- male have under developed testis, tall with long arm development of breast ie. Gynaecomastig and no spermatogenesis & individual are sterile

## 2) Turner's syndrome (X monosomy / XO female

- It is caused due to non-disjunction of chromsome during gamate formation
- Turner's syndrome has 44 Autosome with XO
- They are phenotypically female They have short stature webbed neck, broad shield-shaped chest poorly developed ovaries & breast & low intelligence