

Inheritance and variation

Heredity

"Transfer of genetic character from generation is known as Heredity.

*study of Heredity
is nothing but inheritance.*

* Father of genetics → Gregor Mendel

Seven pairs of contrasting visible characters in pea plant (<i>Pisum sativum</i>)								
Pea Plant Traits								
Dominant	Seed shape	Seed color	Pod shape	Pod color	Flower color	Flower location	Plant height	
	Round	Yellow	Inflated	Green	Purple	Axial	Tall	
Dominant	Round	Yellow	Inflated	Green	Purple	Axial	Tall	
Recessive	Wrinkled	Green	Constricted	Yellow	White	Terminal	Short (Dwarf)	

Reasons for Mendel's Success :

- His experiments were carefully planned and involved large sample.
- He carefully recorded the number of plants of each type and expressed his results as ratios.
- In the pea plant, contrasting characters can be easily recognized.
- The seven different characters in pea plant were controlled by a single factor each. The factors are located on separate chromosomes and these factors are transmitted from generation to generation.
- He introduced the concepts of dominance and recessiveness.

Before learning about Mendel's experiments let us get acquainted with genetic terms and symbols.

Genetic Terminology :-

* Character :-

- It is a specific feature of an organism e.g Height of stem

* Trait :-

- Inherited character and its detectable variants. e.g Tall / Dwarf

* Factor :-

- unit of heredity
- particle present in the organism which is responsible for the inheritance and expression of character.

1st gen → ^{factor} → 2nd gen
 through
 gametes

* genes :-

- particular segment of DNA which is responsible for inheritance and expression of character.

* Allele / Allelomorphs :-

- Two or more alternative form of given genes are called as alleles of each other.
- Allele ← short form of **Allelomorphs**

* Dominant :-

- It is an allele that expresses in the presence of an alternative allele.

* Recessive :-

- This allele is not expressed in the presence of an alternative allele.

* Phenotype :-

- The external appearance of an individual for any traits is called phenotype for that trait.
- It is observable and we can determine it.



* Genotype :-

- genetic constitution or genetics make up of an organism with respect to a particular traits

* Homozygous (pure)

- An individual possessing identical alleles for a particular traits, is called Homozygous or pure for that traits.

* Heterozygous :-

- An individual possessing contrasting alleles for a particular traits, is called as Heterozygous.

* pure line :-

- An individual or a group of individual (population) which is Homozygous or true breeding for one or more traits, constitutes pure line.

* monohybrid :-

- It is Heterozygous for one unit trait and is produced from a cross between two pure parents differing in single pair of contrasting characters e.g. Hybrid tall produced in a cross between pure tall and pure dwarf parents.

* F₁ generation :-

- First filial generation.
- It consists of all off-spring produced from parental cross.

* F₂ generation :-

- The 2nd generation (progeny) produced by selfing (inbreeding) of F₁ generation is called second filial generation.

* Punnett square / checker Board :-

- It is a probability table representing different permutations and combination of fertilization between gametes of the opposite mating types.

* Homologous chromosomes :-

- The morphological, genetically and structurally essentially identical chromosomes present in diploid cell.

* Back cross :-

- It is a cross of F₁ progeny with any of the parents.

* Test cross :-

- It is a cross of F₁ progeny with homozygous recessive parents.

* phenotypic ratio :-

- It is the ratio of the offspring produced in F₂ and subsequent generation with respect to their physical appearance e.g:- STall : I Dwarf

* Genotypic ratio :-

- It is the ratio of the offspring produced in the F₂ and subsequent generation with respect to their genetic make up e.g $1TT : 2Tt$

* monohybrid cross :-

- A cross between parents differing in only one heritable traits is called monohybrid cross.

* Dihybrid cross :-

- A cross between parents differing in two heritable traits, is called dihybrid cross e.g Cross of pure tall, round seeded plant with dwarf, wrinkled seeded plants.

Complete the following chart :

Phenotype	Tall	X	Dwarf										
Genotype	TT		tt										
Gametes	T		t										
First filial Generation (F_1)		Tt											
Selfing of F_1	Tall												
Genotype	Tall	\times	Tall										
Gametes	Tt		Tt										
Second filial Generation (F_2)		<table border="1"> <tr> <td>T</td> <td>t</td> </tr> <tr> <td>Male gametes</td> <td></td> </tr> <tr> <td>Female gametes</td> <td>→</td> </tr> <tr> <td>T</td> <td>Tt</td> </tr> <tr> <td>t</td> <td>Tt</td> </tr> </table>	T	t	Male gametes		Female gametes	→	T	Tt	t	Tt	
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Complete the following chart :

Parental generation

Phenotype	Round, yellow seeds	X	Wrinkled, green seeds
Genotype	RRYY		rryy
Gametes	RY		ry
First filial generation (f_1)		RrYy	
			Round, yellow seeds
Selfing of f_1	Round, yellow seeds	\times	Round, yellow seeds
Genotype	RrYy		RrYy
Gametes	RY, Ry, rY, ry		RY, Ry, rY, ry

mendel laws of inheritance

Law of Dominance

law of Segregation

law of Independent assortment

Statement of Law of Dominance : "When two homozygous individuals with one or more sets of contrasting characters are crossed, the alleles (characters) that appear in F_1 are dominant and those which do not appear in F_1 are recessive".

Statement of Law of Segregation : The law states that "When hybrid (F_1) forms gametes, the alleles segregate from each other and enter in different gametes". The gametes formed are pure in that they carry only one allele each (either dominant allele or recessive allele). Hence, this law is also described as "Law of purity of gametes".

Statement of Law of Independent Assortment: The law states that "When hybrid (F_1) forms gametes, the factors in each pair segregate independently of the other pair".

* Back cross :-

- F_1 individual obtained in a cross usually selfed to get the F_2 progeny.
- they can also be crossed with one of the two parents from which they were derived

* Test cross :-

- the cross of F_1 hybrid with the Homozygous Recessive parents is known as a test cross
- It is used to test whether an individual is Homozygous (pure) or Hetero.

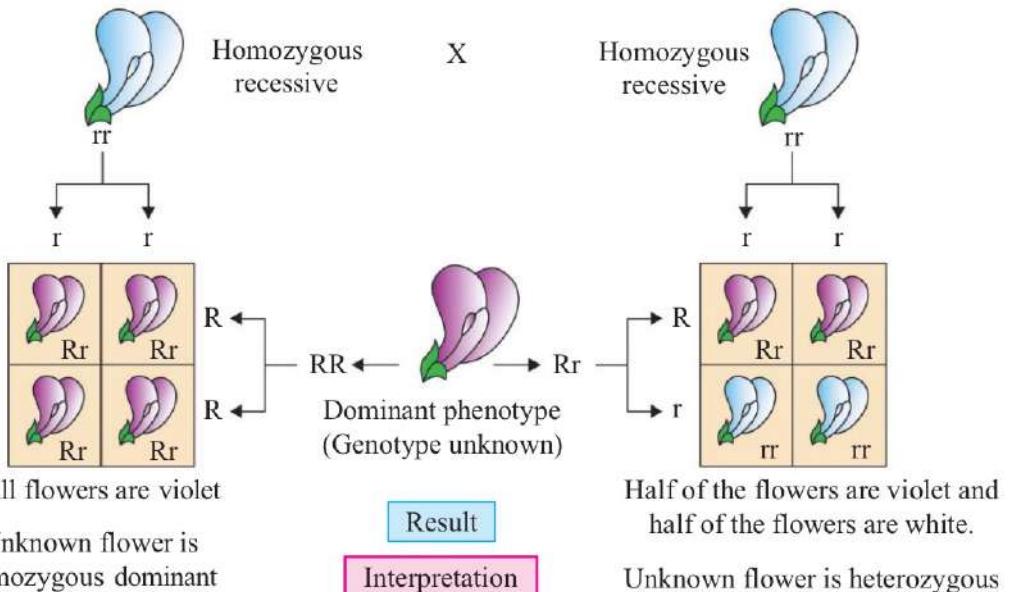


Fig. 3.1 : Graphical representation of test cross

Derivations from Mendel's finding

- Single trait Single gene Two alleles.
- Two alleles show interaction in which one is completely dominant.
- Factors (genes) for different traits present on different chromosomes assort independently.

With the passage of time, number of deviations were observed/ identified in the post-Mendelian era, that gave additional information on the patterns of inheritance. These deviations are then described as Neo-Mendelism.

Note:- phenotypic expression can be modified or influenced by other gene.

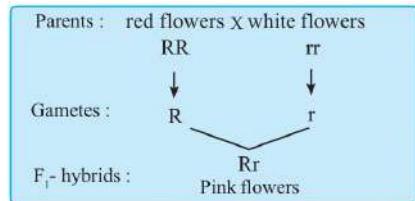
Intragenic
Interaction

Intergenic (non-allelic)
Interaction

- occur between the alleles of same genes
- incomplete dominance and co-dominance
- occur between the alleles of different genes present on the same or different chromosomes
- e.g. pleiotropy, polygenes, epistasis, supplementary and complementary genes.

④ Incomplete dominance :-

- Both the alleles (genes) of an allelomorphic pair express themselves partially.
 - One allele (gene) cannot suppress the expression of the other allele (gene) completely.
- e.g. - *mirabilis jalapa*



F₂ Generation : Selfing of F₁

	R	r
	RR red	Rr pink
R	Rr pink	rr white
r		

Result :

Genotypic ratio - 1RR : 2Rr : 1rr

Phenotypic ratio - 1Red : 2 Pink : 1 White

⑤ Co-dominance :-

- Both the alleles (genes) of an allelomorphic pair express themselves equally in F₁ hybrids.
- Such alleles which are able to express themselves equally independent in hybrid are called co-dominant alleles.
- Both alleles are expressed.

e.g. - coat colours in cattle.

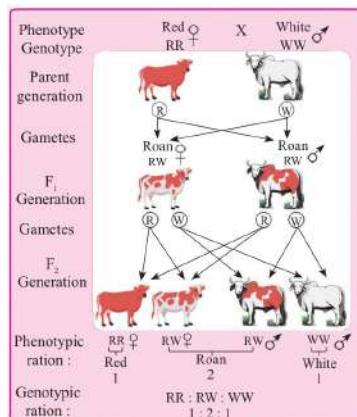


Fig. 3.2 : Representation of co-dominance in cattle

© multiple alleles :-

- more than two alternative forms (alleles) of gene in population occupying the same locus on a chromosome / its Homologue, are known as multiple alleles.
 - multiple alleles arise by mutation of the wild type of gene.
 - A gene can mutate several times producing a series of alternative expression.
- e.g. *Drosophila*

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 or pleiotropism.

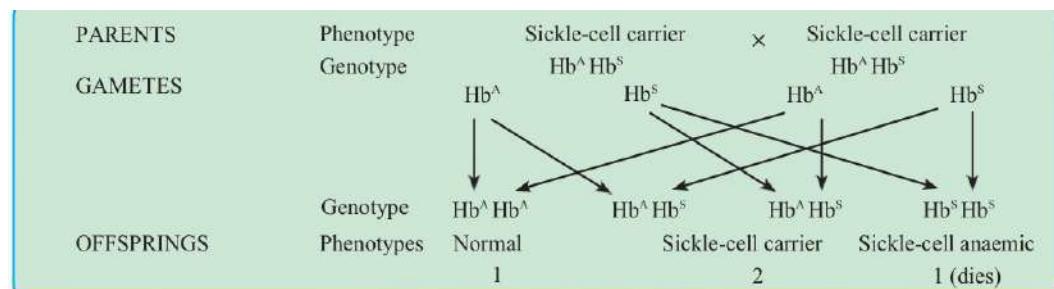


Fig. 3.4 : Representation of Pleiotropy

* chromosomal theory of inheritance :-

Gregor Johann Mendel published his work on inheritance of traits in 1866 but for some reasons, it remained unnoticed or unrecognised till 1900, as communication was not easy in those days. His work was not widely recognized. His approach of using mathematics and statistics to explain biological phenomenon was totally new and unacceptable to the then biologists. As continuous variations were observed in nature, Mendel's concept of factors (genes) as stable and discrete unit which controlled the expression of characters, and that a pair of alleles did not "blend" with each other, was not accepted by his contemporaries. He also did not know the physical location of the 'factors' (genes) in the gametic cell.

In 1900, three scientists Hugo de Vries, Correns and von Tschermark, independently rediscovered Mendel's work on the inheritance of traits. Due to advancements in microscopy, scientists were able to observe cell division and the structure of chromosomes under microscope.

Walter Sutton along with Theodor Boveri (1903) studied the parallel behaviour of Mendel's factors (genes) and behaviour of chromosomes, at the time of meiosis.

Based on these observations, **chromosomal theory of inheritance** was put forth by Sutton and Boveri. This theory identifies chromosomes as the carriers of genetic material.

This theory states that the chromosomes are present in pairs in somatic cells. During gamete formation homologous chromosomes pair, segregate and assort independently during meiosis. Thus, each gamete contains only one chromosome from a pair.

Nucleus of gametes contains chromosomes, which carry all hereditary traits. Male and female gametes (sperms and eggs) carry all the hereditary traits. They are the link between parents and offsprings. The fusion of haploid male gamete and haploid female gamete, restores the diploid number of chromosomes of the species.

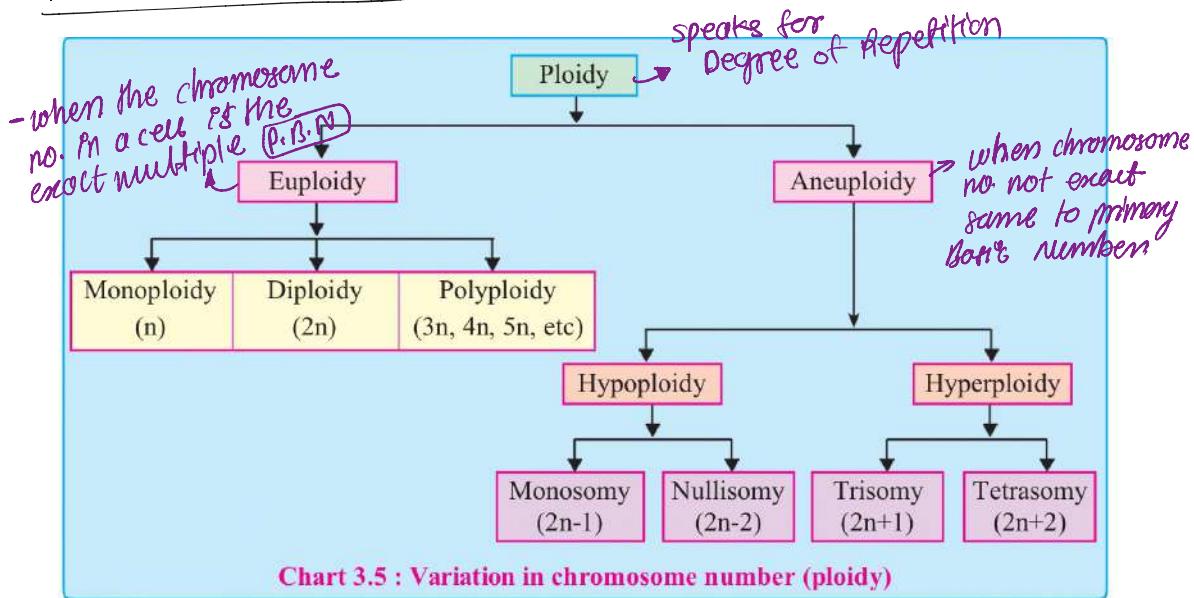
* chromosomes :-

- chromosomes are filamentous bodies present in eukaryotic nucleus
- the term chromosomes (*chromo* = colour, *soma* = body).
- The size of chromosome varies from species to species
- each metaphase chromosome varies from 0.1 - 33 um in length and 0.2 - 2 um in thickness.
- chromosomes are visible during cell division
- they are capable of self replication and play vital role in heredity, mutation, variation and evolutionary development of eukaryotic species.

function :-

- chromosomes are carriers of heredity.

number of chromosomes :-



structure of chromosome :-

- chromosome are best visible under microscope, when the cell is at metaphase stage.
- ④ because chromosomes are highly condensed.
- typical chromosome contains two chromatids joined together at centromere / primary constriction.

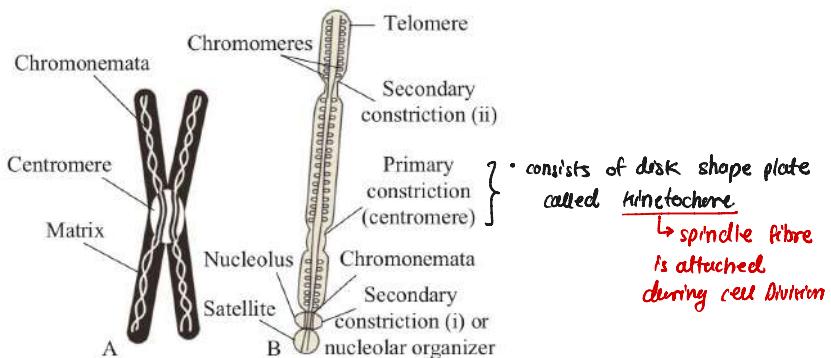


Fig. 3.6 : Structure of Chromosome

Sex chromosome :- (Allosomes)

- chromosome which are responsible for the determination of sex.

- Human Being $\begin{cases} X \\ Y \end{cases}$

④ X-chromosome

- chromosome is straight, rod like and longer than Y chromosome.

- metacentric.

- Has large amount of chromatin and small amount of Heterochromatin

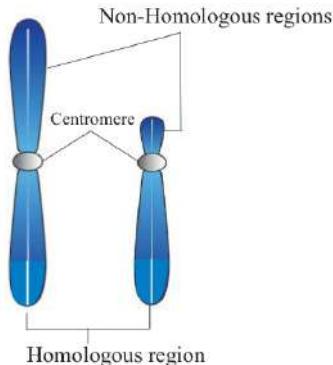
⑤ Y-chromosome :-

- smaller than X-chromosome

- acrocentric

- Y chromosome has small amount of euchromatin and large amount of Heterochromatin.

- genetically less active/inert.



: Structure of X and Y chromosomes
(in humans)

* Linkage :-

- It is known fact that several genes are present on the chromosome
- As chromosome are carriers of heredity, these genes have tendency to be inherited together.
- such genes are called linked genes.

"This tendency of two or more genes present on the same chromosome that are inherited together is known as linkage."

I. Complete linkage : The linked genes which are closely located on the chromosome do not separate (no crossing over) and inherit together. They are called completely linked (strongly linked) genes and the phenomenon of their inheritance is called complete linkage. Thus the parental traits are inherited in offsprings. e.g. X chromosome of *Drosophila* males- show complete linkage.

II. Incomplete linkage : The linked genes which are distantly located on the same chromosome and have chances of separation by crossing over, are known as incompletely linked (weakly linked) genes. The phenomenon of their inheritance, is called incomplete linkage. Thus, new traits occur in offsprings. e.g. In *Zea mays* - colour and shape of grain show incomplete linkage.

* Linkage groups :-

"All the linked genes in a particular chromosomes, constitute a linkage group."

for eg \Rightarrow *Drosophila melanogaster* has 4-linkage groups that correspond to the 4-pairs of chromosomes.

* Sex-linkage :-

"The transmission (inheritance) of x-linked and y-linked genes from parents to offspring , called sex-linked inheritance.

sex-linked inheritance :-

- X-linked
- Y-linked
- XY-linked

a. 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.

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

b. 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. Examples of X-Y linked traits are total colour blindness, nephritis, retinitis pigmentosa, etc.

Crossing Over :

Crossing over is a process that produces new combinations (recombinations) of genes by interchanging and exchanging of corresponding segments between non-sister chromatids of homologous chromosomes. It occurs during pachytene of prophase I of meiosis. The term crossing over was coined by Morgan. The mechanism of crossing over consists four sequential steps such as synapsis, tetrad formation, crossing over and terminalization. This you have already studied in the chapter on cell division in class XI. The phenomenon of crossing over is universal and it is necessary for the natural selection, because it increases the chances of variation.

3.9 Autosomal Inheritance :

Human somatic ($2n$) cell contains

23 pairs of chromosomes. They can be divided functionally as autosomes and sex chromosomes. A single pair of chromosomes 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 example,

- Autosomal dominant traits like Widow's peak and Huntington's disease, etc.
- Autosomal recessive traits like Phenyl ketonuria (PKU), Cystic fibrosis and Sickle cell anaemia.

a. Widow's peak :

A prominent "V" shaped hairline on forehead is described as widow's 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 widow's peak). Both males and females have equal chance of inheritance.

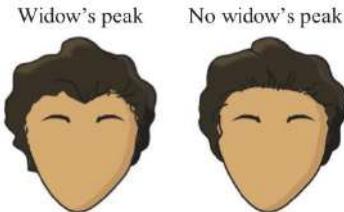


Fig. 3.9 : Widow's peak and straight hair line

b. Phenylketonuria (PKU):-

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 and cerebrospinal fluid (CSF). It affects development of brain and causes mental retardation. Excess phenylalanine is excreted in urine, hence this disease is called phenylketonuria.

sex linked inheritance :-

- genes located on non-homologous region of sex chromosome, are called sex-linked genes.
- The traits that are determined by sex linked genes, are called sex-linked traits.

a. X-linked (sex linked) genes :

The X linked genes are located on non homologous region of X chromosome and these gene do not have corresponding alleles on Y chromosome.

Female has two X chromosomes. In female two recessive sex linked genes are required for expression of sex linked traits. If one X chromosome carries a recessive gene for sex-linked trait (defect) its effect is suppressed by the dominant gene present on other X chromosome. The females with one recessive gene are carriers. The carrier female is physically normal as she does not suffer from the disease (disorder).

Male has only one X-chromosome. If X chromosome carries X-linked recessive gene for sex linked trait, then it is expressed phenotypically, because there is no dominant gene on Y chromosome to suppress its effect. Therefore, sex-linked / X-linked traits appear more frequently in males than in the females. Examples of X-linked traits include haemophilia, colour blindness, night blindness, myopia, muscular dystrophy, etc.

b. Y-linked (Holandric) genes :

Genes located on non-homologous region of Y chromosome, are called Y linked genes. The Y-linked genes are inherited directly from male to male. In man, the Y-linked genes such as hypertrichosis is responsible for excessive development of hair on pinna of ear. This character is transmitted directly from father to son.

Colour blindness :

Colour blindness is X-linked recessive disorder where person is unable to distinguish between red and green colours as both the colours appear grey. It is caused due to recessive X-linked genes (X^c) which prevents formation of colour sensitive cells, the cones, in the retina of eye.

The homozygous recessive females ($X^c X^c$) and hemizygous recessive male ($X^c Y$) are unable to distinguish between red and green colours. The frequency of colour blind women is much less than colour blind men. Dominant X linked gene (X^C) is necessary for formation of colour sensitive cells in the retina of eye. Thus, genotypes of male and female individuals can be represented as follows-

Sex	Normal	Colourblind	Carrier
Male	$X^C Y$	$X^c Y$	—
Female	$X^C X^C$	$X^c X^c$	$X^C X^c$

The inheritance of colourblindness 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 F_1 . The sons have normal vision but daughter will be carrier for the disease.

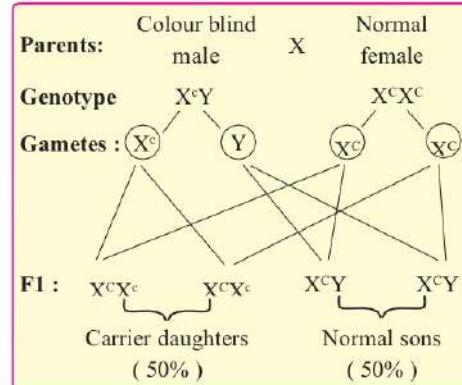


Fig. 3.10 : Sex linked inheritance (colour blindness)

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

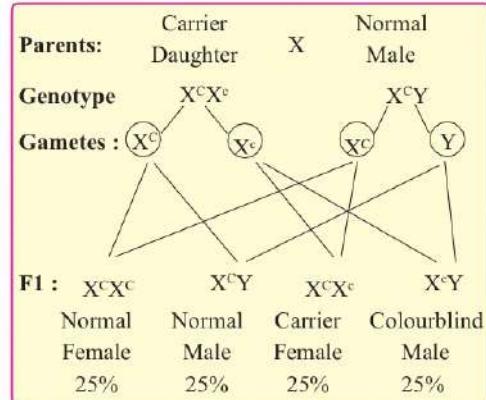


Fig. 3.11 : Sex linked inheritance (colour blindness)

From above example, it is clear that the X linked recessive gene for colour blindness is inherited from colourblind father to his grandson through his daughter. This type of inheritance is called as cris-cross inheritance.

Haemophilia (Bleeder's disease) :

Haemophilia is X-linked recessive disorder in which blood fails to clot or coagulates very slowly. The 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. Even minor injuries cause continuous bleeding, hence haemophilia is also called as bleeder's disease.

The recessive gene for haemophilia is located on non homologous region of X chromosome. As there is no corresponding allele on Y chromosome to suppress its expression, so men suffer from this disease. Women suffers only when both X chromosomes have recessive genes (alleles).

The genotype of male and female individuals can be represented as follow-

Sex	Normal	Haemophilic	Carrier
Male	$X^H Y$	$X^h Y$	—
Female	$X^H X^H$	$X^h X^h$	$X^H X^h$

Like colour blindness, haemophilia also shows criss-cross inheritance. The inheritance of haemophilia can be studied with the help of following examples -

1. Marriage between the Haemophilic male and normal female.

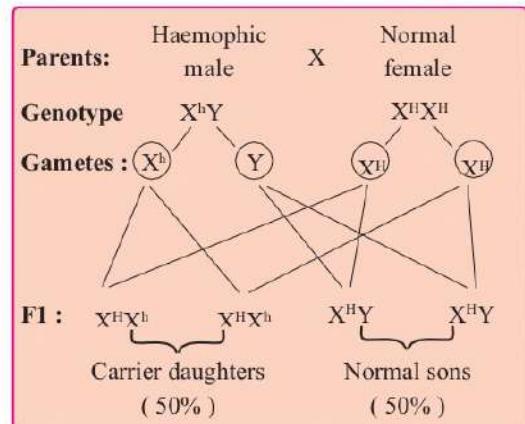


Fig. 3.12 : Sex linked inheritance

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

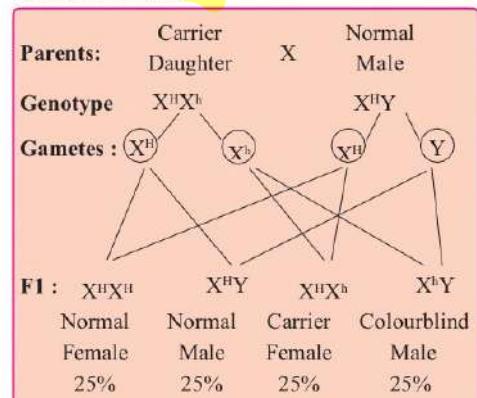


Fig. 3.13 : Sex linked inheritance
(Haemophilia)

3.11 Sex Determination :

The mechanism by which sex is established is termed as **sex determination**. The term sex refers to sexual phenotype. In some species, both male and female reproductive organs are present in same organism. It is described as **bisexual or hermaphrodite or monoecious**.

a. Sex Determination in human beings :

The chromosomal mechanism of sex determination in human beings is XX-XY type. In human beings, the nucleus of each somatic cell contains 46 chromosomes or 23 pairs of chromosomes. Out of these, 22 pairs are **autosomes** and one pair of **sex chromosomes**.

Human female has a pair of XX, homomorphic sex chromosomes while male has XY, heteromorphic sex chromosomes.

Thus genotype of :

Female = 44 Autosomes + XX

Male = 44 Autosomes + XY

During gamete formation in male, the diploid germ cells in testis undergo spermatogenesis to produce two types of haploid sperms, 50% sperms contain 22 autosomes and X chromosome while, 50% sperms contain 22 autosomes and Y chromosome.

In Female, the diploid germ cells in ovaries undergo oogenesis to produce only one type of egg. All eggs contain 22 autosomes and X chromosome. Thus human male is heterogametic and female is homogametic.

If sperm containing X chromosome fertilizes egg (ovum), then diploid zygote is formed, that grows into a female child. If

On the other hand, some species in which the organism has either male or female reproductive organs, is said to be **dioecious** or **unisexual**. Humans are dioecious.

German biologist, Henking in 1891, while studying spermatogenesis of the squash bug (*Anasa tristis*), noted that 50% of sperms receive the unpaired chromosomes while other 50% sperm do not receive it. Henking gave a name to this structure as the x-body but he could not explain its role in sex determination. Further investigations by other scientists led to conclusion that the "x-body" of Henking was in fact a chromosome and gave the name 'X-Chromosome'.

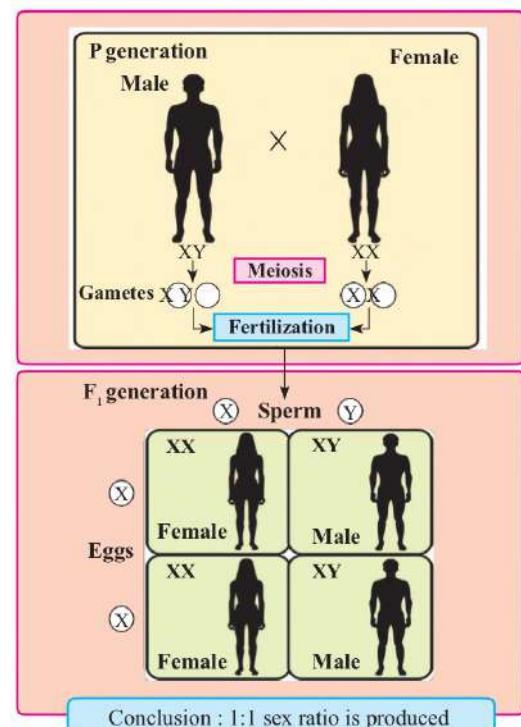


Fig. 3.14 : Sex determination in human beings

b. Sex Determination in birds :

In birds, the chromosomal mechanism of sex determination is ZW-ZZ type. In this type females are heterogametic and produce two types of eggs; 50% eggs carry Z- chromosome, while 50% eggs carry W- chromosome.

Males are homogametic and produce one type of sperms. Each sperm carries a Z-chromosome. Thus sex of individual depends on the kind of egg (ova) fertilized by the sperm.

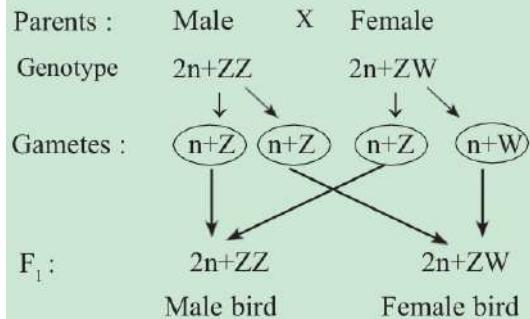


Fig. 3.15 : Sex determination in birds

c. Sex Determination in honey bees :

In honey bees, chromosomal mechanism of sex determination is **haplo-diploid type**. In this type, sex of individual is determined by the number of set of chromosomes received. Females are diploid ($2n=32$) and males are haploid ($n=16$). The female produces haploid eggs ($n=16$) by meiosis and male produces haploid sperms ($n=16$) by mitosis. If the egg is fertilized by sperm, the zygote develops into

a diploid female ($2n=32$) (queen and worker) and unfertilised egg develops into haploid male ($n=16$) (Drone) by way of parthenogenesis.

The diploid female gets differentiated into either worker or queen depending on the food they consume during their development. Diploid larvae which get royal jelly as food develops into queen (fertile female) and other develops into workers (sterile females).

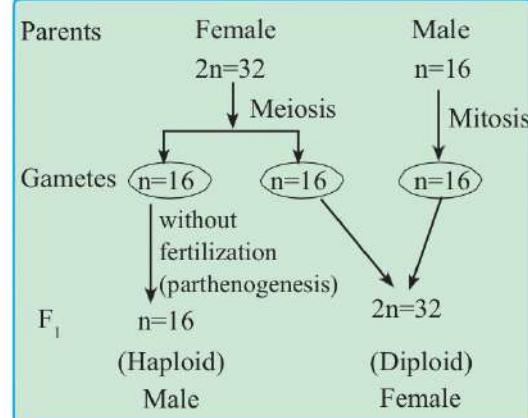


Fig. 3.16 : Sex determination in honey bee

3.12 Genetic Disorders :

Genetic Disorders are broadly grouped into two categories as, **Mendelian disorders** and **chromosomal disorders**. Mendelian disorders are mainly caused due to alteration or mutation in the gene. e.g. thalassemia, sickle-cell anaemia, colourblindness, haemophilia, phenylketonuria, etc. On the other hand, chromosomal disorders are caused due to absence or excess of one or more chromosomes or their abnormal arrangement. For eg, Down's syndrome, Turner's syndrome, Klinefelter's syndrome etc.

Thalassemia :

Thalassemia is an autosomal, inherited recessive disease. Haemoglobin molecule is made of four polypeptide chains- 2 alpha (α) and 2 beta (β) chains. The synthesis of alpha chains are controlled by two closely linked genes (HBA1 and HBA2) on chromosome 16 while the synthesis of beta chain is controlled by a single gene (HBB) on chromosome 11.

Depending upon which chain of haemoglobin is affected, thalassemia is classified as alpha-thalassemia and beta-thalassemia. It is caused due to deletion or mutation of gene which codes for alpha (α) and beta (β) globin chains that result in abnormal synthesis of haemoglobin. In Thalassemia, person shows symptoms like anaemia, pale yellow skin, change in size and shape of RBCs, slow growth and development, dark urine, etc.

Down's Syndrome (21st trisomy) :

Down's syndrome is named after the physician John Langdon Down who first described this autosomal chromosomal disorder in 1866.



Fig. 3.17 : Down's Syndrome

This Syndrome is caused due to an extra copy of chromosome number 21st. It shows presence of three copies of 21st chromosome instead of homologous pair. These individuals will have 47 chromosomes instead of the normal number 46. 21st Trisomy occurs due to non-disjunction or failure of separation of chromosomes (**autosomes**) during gamete formation. The incidence of non-disjunction is distinctly higher in mothers who are over 45 years old.

These patients have mild or moderate mental retardation and skeletal development is poor. Distinct 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, flat hands and stubby fingers and palm is broad with single palmar crease.

Turner's Syndrome :

(X monosomy / XO females)

It is sex chromosomal disorder caused due to non-disjunction of chromosome during gamete formation. Individual born with Turner's syndrome has 44 autosomes with XO. They are phenotypically female. They have a short stature (height) and webbed neck, lower posterior hair line, broad shield-shaped chest, poorly developed ovaries and breast, and low intelligence.

Down Syndrome - Trisomy 21

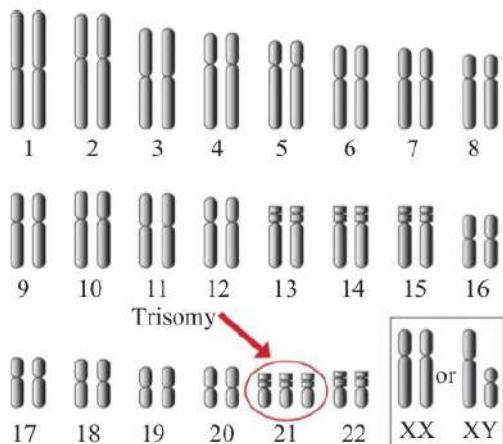


Fig. 3.18 : Karyotype of Down's syndrome

Klinefelter's syndrome (XXY males) :

It is chromosomal disorder caused due to extra X chromosome in males. Thus genotype of individuals is 44 + XXY. They are described as feminized males. Extra chromosome is a result of non-disjunction of X-chromosome during meiosis. Individual is male and has over all masculine development. Voice pitch is harsh and have under developed testis. They are tall with long arms, feminine development (development of breast i.e. Gynaecomastia) and no spermatogenesis, therefore, individuals are sterile.