3 Inheritance and Variation

Can you recall?

1. Is there a similarity between the parents and offsprings?
2. What are the roles of chromosomes in living organisms?
3. How are hybrid seeds produced?
4. Which are the chromosomal disorders?

3.1 Chromosomes and Mechanism of inheritance.

- The transmission of genetic information from generation to generation is known as **heredity** or **inheritance**. The mechanism of inheritance was successfuly investigated before chromosomes had been observed or genes were known.
- Gregor Mendel, son of the peasant farmer, was born in Moravia in 1822. Gregor Mendel first gave the accurate explanation for the mechanism of inheritance by using hybridization technique.
- Inheritance of the seven traits in garden pea (Refer the diagram below) plant were studied individually one at a time or in combination of two or three character at a time. He processed the data mathematically and statistically.
- Mendel postulated the principles of heredity which then became the fundamental laws of heredity, as proposed by Correns (1900). He visualized that the traits as such are not inherited physically but by 'something' present inside the gametic cell. To this 'something', he coined term 'factors' that are responsible for expression of a particular trait/ character. He proposed that factors are particulate in nature. The Mendelian factors are now termed as 'genes'. These factors occur in pairs in the parents and segregate from each other during gamete formation without blending/ mixing.

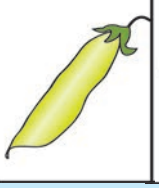
Reasons for Mendel's Success :

- 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.

Do you know ?

Seven pairs of contransting visible characters in pea plant (*Pisum sativum*)

Pea Plant Traits

	Seed shape	Seed color	Pod shape	Pod color	Flower color	Flower location	Plant height
Dominant	Round 	Yellow 	Inflated 	Green 	Purple 	Axial 	Tall 
Recessive	Wrinkled 	Green 	Constricted 	Yellow 	White 	Terminal 	Short (Dwarf) 

- In the pea plant, contrasting characters can be easily recognized.
- The seven different charaters 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.

3.2 Genetic Terminology :

Character : It is a specific feature of an organism e.g. height of stem.

Trait : An inherited character and its detectable variant e.g. Tall or dwarf.

Factor : It is a unit of heredity, a particle present in the organism which is responsible for the inheritance and expression of a character. (factor is passed from one generation to the next through gametes). Factor determines a genetical (biological) character of an organism.

Gene : It is a particular segment of DNA which is responsible for the inheritance and expression of that character.

Alleles or Allelomorphs : The two or more alternative forms of a given gene (factor) are called alleles of each other. They occupy identical loci (positions) on homologous chromosomes. Allele is a short form of Allelomorph.

Dominant : It is an allele that expresses its trait even in the presence of an alternative allele i.e. in heterozygous condition only. Alternatively, the allele that expresses in F_1 is called dominant. (It is an allele of a pair that masks the expression of other allele in F_1 generation.)

Recessive : This allele is not expressed in the presence of an alternative allele (in heterozygous condition). It expresses only in the presence of another identical allele. It is an allele that does not express in F_1 hybrid.

Phenotype : The external apperance of an individual for any trait is called phenotype for that trait. It is observable and is determined by different combinations of alleles. e.g. In pea, for the height of stem (plant) tall and dwarf are the two phenotypes (Tall is determined by TT or Tt and dwarf by tt).

Genotype : Genetic constitution or genetic make up of an organism with respect to a particular trait. It is representation of the genetic constitution of an individual with respect to a single character or a set of characters. e.g. pea tall plants can have genotype TT or Tt and dwarf has tt.

Homozygous (pure) : An individual possessing identical allels for a particular trait, is called homozygous or pure for that trait.

Homozygous breeds true to the trait and produces only one type of gametes e.g., tall with TT and dwarf with tt.

Heterozygous : An individual possessing contrasting alleles for a particular trait, is called heterozygous. Heterozygous does not breed true for that trait and produces two types of gametes e.g. F_1 generation hybrids (Tt). Heterozygous individual is also called hybrid.

Pure line : An individual or a group of individuals (population) which is homozygous or true breeding for one or more traits, constitutes pure line i.e. plant which breeds true for a particular character. It is a descendent of a single homozygous parent produced after self fertilization.

Monohybrid : It is heterozygous for one 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. It is a heterozygote for a single pair of alleles.

F_1 generation : It refers to the first filial generation. It consists of all off-springs produced from a parental cross. Alternatively, it is first generation from a given mating between pure parents having contrasting characters.

F_2 generation : The second generation (progeny) produced by selfing (inbreeding) of F_1 generation offsprings is called second filial generation. e.g. Progeny produced from a cross between two F_1 individuals (e.g. Tt × Tt).

Punnett square/checker board : It is a probability table representing different permutations and combination of fertilization between gametes of the opposite mating types. In short, it is a diagrammatic representation of a particular cross to predict the progeny of a cross.

Homologous Chromosomes : The morphologically, genetically and structurally essentially identical chromosomes present in a diploid cell, are called homologous chromosomes. Such chromosomes synapse during meiosis.

Back cross : It is a cross of F_1 progeny with any of the parents (e.g. F_1 tall × pure tall; F_1 tall × pure dwarf i.e. Tt × TT/tt).

Test cross : It is a cross of F_1 progeny with homozygous recessive parent (e.g. F_1 tall × pure dwarf i.e. Tt × tt). It is used to test the homozygous/ heterozygous nature of hybrid. It is a kind of back cross.

Phenotypic ratio : It is the ratio of the offsprings produced in F_2 and subsequent generation with respect to their physical appearence e.g. 3Tall : 1 dwarf, is F_2 'Phenotypic ratio' in monohybrid cross.

Genotypic ratio : It is the ratio of the offsprings produced in the F_2 and subsequent generation with respect to their genetic make up e.g. 1 TT : 2Tt : 1 tt, is F_2 genotypic ratio in monohybrid cross.

Monohybrid cross :

A cross between parents differeing in only one heritable trait is called monohybrid cross. e.g. cross of pure tall and pure dwarf plants. Mendel performed the monohybrid cross between two pea plants with only one pair of contransting character.

Activity :

Complete the following chart :

Phenotype	Tall	X	Dwarf
Genotype	TT		☐
Gametes	T		t
First filial Generation (F_1)	Tt Tall		
Selfing of F_1	Tall	×	Tall
Genotype	Tt		Tt
Gametes	☐		☐

Second filial Generation (F_2)

Female gametes ↓ / Male gametes →	T	t
T		
t		

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 plant. Mendel also performed the dihybrid cross between pea plants that differed in two pairs of contrasting characters.

Activity :

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	×	Round, yellow seeds
Genotype	RrYy		RrYy
Gametes	RY, Ry, rY, ry		RY, Ry, rY, ry

F_2 Generation :

Male gametes → / Female gametes ↓	RY	Ry	rY	ry
RY				
Ry				
rY				
ry				

Use your brain power

There are 16 possible individuals in F_2 generation. Try to find out the phenotypes as well as the genotypic and phenotypic ratios.

Can you tell?

Why are farmers and gardeners advised to buy new F_1 hybrid seeds every year?

3.3 Mendel's Laws of Inheritance :

Mendel proposed three basic postulates on the basis of which three laws were formulated. These are described below:

1. Law of Dominance :

In monohybrid and dihybrid 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 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**".

2. Law of segregation (Law of purity of gametes) :

This law is based on the fact that the alleles do not show any blending/ mixing and both the 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 (trait). A gamete may carry either dominant or recessive factor but not both. That's why it is also called as law of purity of gametes.

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**".

3. Law of Independent Assortment :

This law is based on dihybrid cross. It is basic principle of genetics developed by a Mendel. It describes how different genes or alleles present on separate chromosomes independently separate from each other, during formation of gametes. These alleles are then randomly united in fertilization. In dihybrid cross, F_2 phenotypic ratio 9:3:3:1 indicates that the two pairs of characters behave independent of each other. It can be concluded that the two characters under consideration are assorted independently giving rise to different combinations.

Statement of Law of Independent Assortment: The law states that "When hybrid possessing two (or more) pairs of contrasting factors (alleles) forms gametes, the factors in each pair segregate independently of the other pair".

Try This

Find the ratio of dihybrid test cross by using punnett square.

3.4 Back Cross and Test Cross :

a. 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**.

b. 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.

Test cross can be used to find out genotype of any plant with dominant expression. But it is not known whether it is homozygous (pure) or heterozygous for that trait. For example, A pea plant having violet (purple) flowers is crossed with a pea plant with white flowers. If all flowers produced are violet, we can conclude that plant is pure or homozygous and if we get violet and white flowers in 1:1 ratio, we can conclude that plant is heterozygous. Test cross is also used to introduce useful recessive traits in the hybrids of self pollinated plants during rapid crop improvement programs.

Following is the graphic representation of test cross (Fig. 3.1). Recessive parent is crossed to find out unknown genotype.

3.5 Deviations from Mendel's findings :

Few generalizations were arrived at by Mendel, on the basis of his experiments of garden pea plant- such as,

i. Single trait ⟶ Single gene ⟶ Two alleles.
ii. Two alleles show interaction in which one is completely dominant.
iii. 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**.

Internet my friend

Find out the principle involved in heredity of sheep/ coat colour

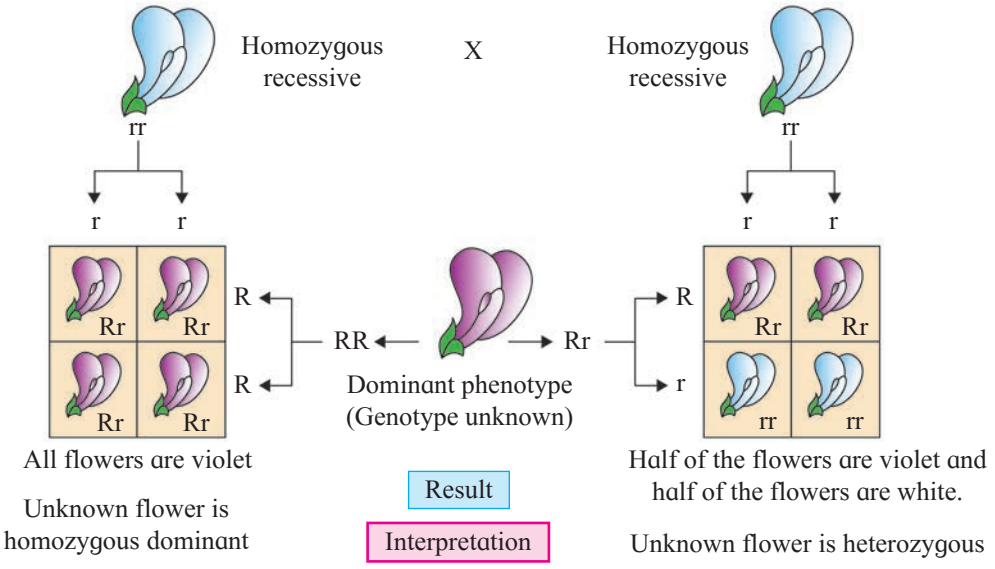


Fig. 3.1 : Graphical representation of test cross

Do you know ?

The deviations are :

a. Single trait ⟶ Single gene ⟶ two alleles showing interactions like codominance and incomplete dominance.

b. Single trait ⟶ Single gene ⟶ more than two alleles (multiple allelism).

c. Single trait ⟶ more than one genes (Polygenic inheritance) showing different epistatic interactions or additive effect.

d. Single gene influencing many traits (pleiotropy).

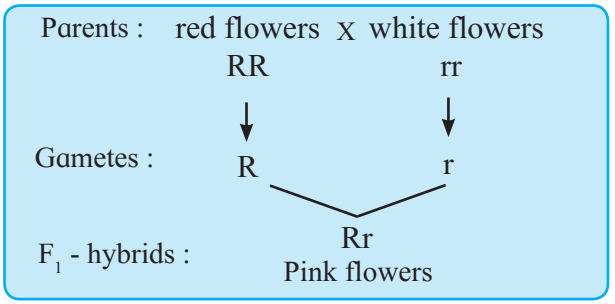
It was observed that the phenotypic expression of a gene can be modified or influenced by the other gene. These gene interactions are of two types.

i. **Intragenic interactions :** Occur between the alleles of same gene e.g. incomplete dominance and co-dominance. It also occurs between the multiple allele series of a gene.

ii. **Intergenic (non-allelic) interactions:** Occur between the allels of different-genes present on the same or different-chromosomes. e.g. pleiotropy, polygenes, epistasis, supplermetary and complementry genes, etc. Some of these interactions are discussed below :

a. Incomplete dominance:

In the incomplete dominance, both the alleles (genes) of an allelomorphic pair express themselves partially. One allele (gene) cannot supress the expression of the other allele (gene) completely. In such case, there is an intermediate expression in the F_1 hybrid. A well-known example is the flower colour of *Mirabilis jalapa*. If a red-flowered (RR) plant is crossed with a white-flowered (rr) plant, then F_1 offsprings have pink (Rr) flowers.



F_2 Generation : Selfing of F_1

♀ \ ♂	R	r
R	RR red	Rr pink
r	Rr pink	rr white

Result :

Genotypic ratio - 1RR : 2Rr : 1rr

Phenotypic ratio - 1Red : 2 Pink :1 White

b. Co-dominance :

In co-dominance, both the alleles (genes) of an allelomorphic pair express themselves equally in F_1 hybrids. Such alleles which are able to express themselves equally independently in hybrids, are called co-dominant alleles. Thus in co-dominance both alleles are expressed.

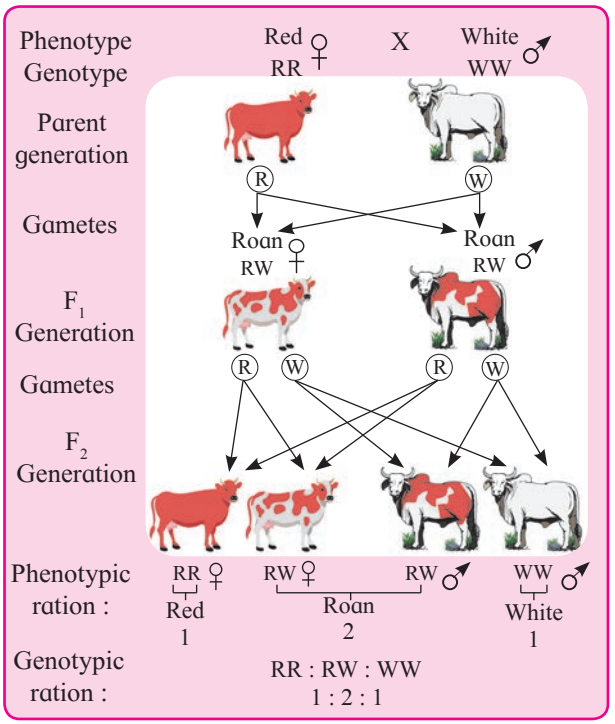


Fig. 3.2 : Representation of co-dominance in cattle

Classic example of co-dominance is coat colour in cattle. There are two types one with red coat (with red colour hair) and other with white coat (with white hair). When red cattles (RR) are crossed with white cattles (WW), F_1 hybrids (RW) are roan.

Roans have the mixture of red and white colour hair. Thus both the traits are expressed equally. In F_2 generation red (RR), roans (RW) and white (WW) are produced in the ratio 1:2:1. Thus in Co-dominance, the genotypic and phenotypic ratios are identical.

c. Multiple alleles :

More than two alternative forms (alleles) of a gene in a population accupying the same locus on a chromosome or its homologue, are known as **multiple alleles**. Multiple alleles arise by mutations of the wild type of gene. A gene can mutate several times producing a series of alternative expression. Different alleles in a series show dominant- recessive relation or may show co-dominance or incomplete dominance among themselves. Wild type is dominant over all other mutant alleles.

In *Drosophila*, a large number of multiple alleles are known. e.g. The size of wings from normal wings to vestigial (no) wings, i.e., just stumps, is due to one allele (vg) in homozygous condition. The normal wing is wild type while vestigial wing 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

Curiosity Box

1. What is qualitative and quantitative inheritance?
2. Find out the traits of quantitative inheritance in humans.

Another good example of multiple alleles is A, B, O blood grouping in human beings.

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**. The phenotypic ratio is 1:2 instead of 3:1 because of the death of recessive homozygote. The disease, sickle-cell anaemia, is caused by a gene Hb^s. Normal or healthy gene Hb^A is dominant. The carriers (heterozygotes Hb^A/Hb^s) show signs of mild anaemia as their RBCs become sickle-shaped i.e. half- moon- shaped only under abnormally low O_2 concentration.

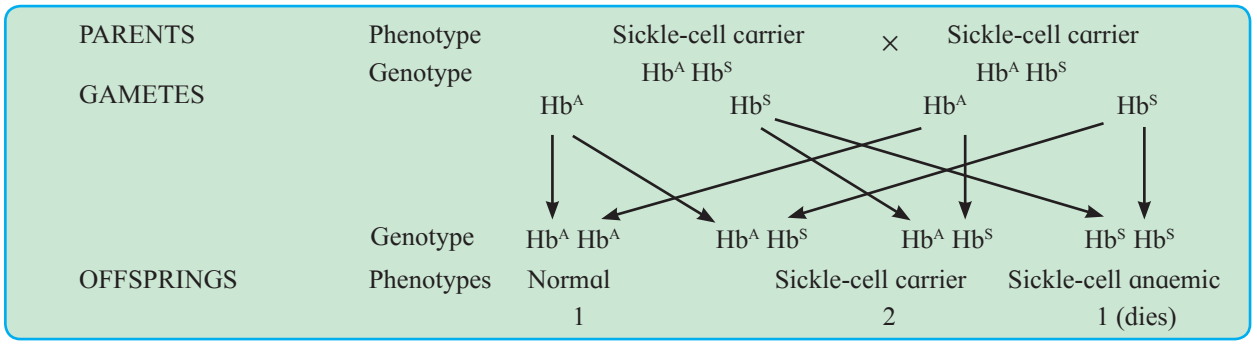


Fig. 3.4 : Representation of Pleiotropy

The homozygotes with recessive gene Hb^s however, die of total anaemia. Thus, the gene for sickle- cell anaemia is lethal in homozygous condition and produces sickle cell trait in heterozygous carrier. Two different expressions are produced by a single gene.

A marriage between two carriers will produce normal, carriers and sickle-cell anaemic children in 1:2:1 ratio. Sickle cell anaemics die leaving carriers and normals in the ratio 1:2. The heterozygotes or carriers can be identified by microscopic examination of blood.

Internet my friend

1. Find out the inheritance pattern in blood groups of human beings.
2. Find out more about pleiotropy-(Sickle-cell anaemia) and Polygenic inheritance - (human skin colour)

3.6 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 Tschermak, independently rediscovered Mendel's work on the inheritance of traits. Due to advancements in microcopy, 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.

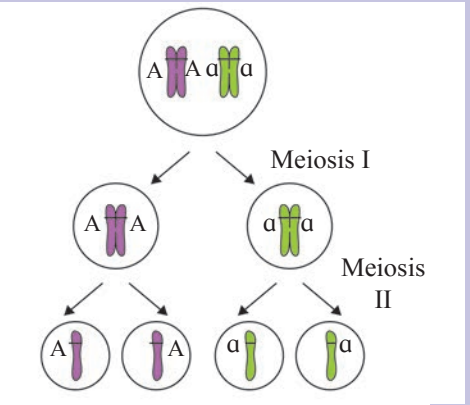
Based on these observations, **chromosomal theory of inheritance** was put forth by Sutton and Boveri. This theory identifies chromosomes as the carriers of genetic material.

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 haplaid female gamete, restores the diploid number of chromosomes of the species.

Activity :

Observe the following diagram and answer the questions given below -



Questions :

1. What is homologous chromosome?
2. In which phase of meiosis-I, homologous chromosomes segregate?
3. Where are genes located?
4. Do genes and chromosomes have similar behaviour? Justify.

Always Remember

1. Genes and chromosomes occur always in pairs in diploid organism.
2. Alleles located on chromosome segregate along with chromosome during gamete formation.

Can you recall?

1. What is chromosome?
2. How many chromosomes are present in human somatic and reproductive cell?

3.7 Chromosomes :

Chromosomes are filamentous bodies present in the eukaryotic nucleus. The term chromosomes (Gr., Chromo = colour, soma = body) was coined by W. Waldeyer (1888). The size of chromosome varies from species to species. Each metaphase chromosome varies from 0.1 to 33 µm in length and 0.2 to 2 µm in thickness. Chromosomes are visible during cell division. They are capable of self replication and play vital role in heredity, mutation, variation, and evolutionary devlopment of eukaryotic species. Chemically eukaryotic chromosomes are made of DNA, histone and non-histone proteins.

Function :

Chromosomes are carriers of heredity.

Number of chromosomes :

The number of chromosomes is specific and constant for a particular species, therefore it is of great importance in the study of phylogeny and taxonomy of the species.

The term **Ploidy** speaks for the degree of repetition of the primary basic number of chromosomes (i.e. 'x') in a cell. When the chromosome number in a cell is the exact multiple of the primary basic number, then it is called **euploidy**. Euploids include monoploid/ haploid (with one set of chromosomes where x=n), diploids (2n-two sets of chromosomes), triploids (3n-three sets of chromosomes), tetraploid (4n-four sets of chromosomes) and so on. When the chromosome number is not the exact multiple of the haploid set, it is described as **Aneuploidy**. Aneuploidy is either addition or deletion of one or more chromosome (s) to the total number of chromosomes in a cell (see the chart 3.5).

Structure of chromosome :

Chromosomes are best visible under microscope, when the cell is at metaphase stage. It is because at this stage chromosomes are highly condensed. Typical chromosome

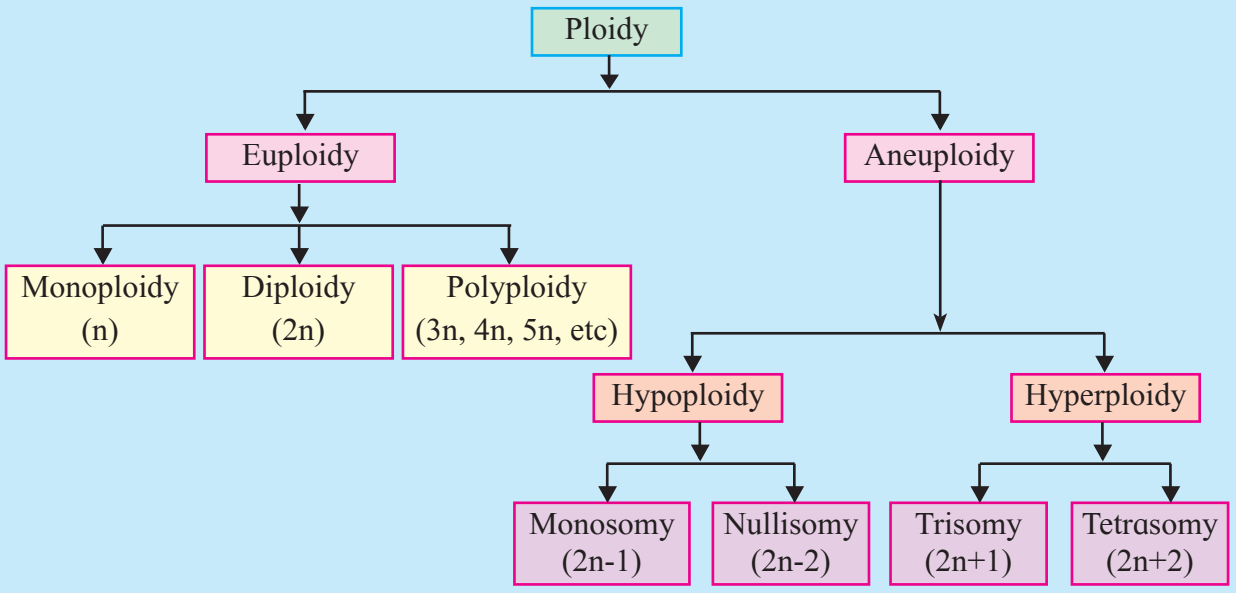
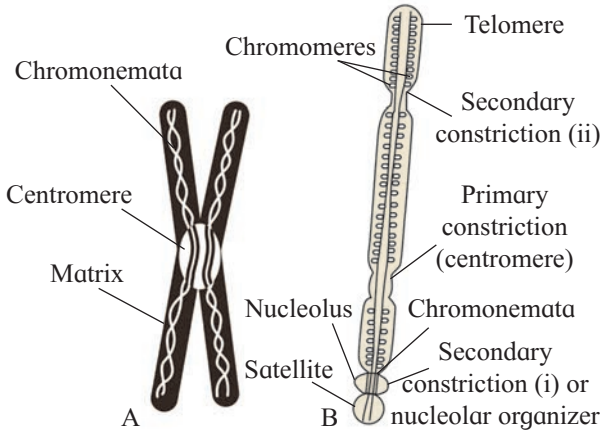


Chart 3.5 : Variation in chromosome number (ploidy)

consists of two chromatides joind together at centromere or primary constriction. Primary constriction constists of a disk shape plate called **kinetochore**. It is at the kinetochore, spindle fibres get attached during cell division. Besides primary constriction, some few chromosomes possess additional one or two constrictions called **secondary constriction**. At secondary constriction I, nucleolus becomes organized during interphase. A satellite body (SAT body) is attached at secondary constriction II, in very few chromosomes. Each chromatid in turn contains a long, unbranched, slender, highly coiled DNA thread, called Chromonema, extending through the length of chromatid.

Chromatid consists a double stranded DNA molecule which extends from one end of chromosomes to other.



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

Fig. 3.6 : Structure of Chromosome

Activity :

Study the types of chromosome according to position of centromere. Observe and complete the following table.

Types of Chromosome	Name of Chromosome	Position of Contromere
	Metacentric	-
	-	-
	Acrocentric	-
	-	At one end

Depending upon the position of centromere there are four types (shapes) of chromosomes viz. Acrocentric (j shaped), Telocentric (i shaped), Submetacentric (L shaped) and Metacentric (V shaped). The ends of chromosome (i.e. chromatids) are known as **telomeres**.

Sex Chromosomes :

The chromosomes which are responsible for the determination of sex are known as **sex chromosomes** (Allosomes). Human being and other mammals have X and Y Chromosomes as sex chromosomes.

X chromosome is straight, rod like and longer than Y chromosome. X chromosome is metacentric, while Y chromosome is acrocentric. X chromosome has large amount of **euchromatin** (extended region) and small amount of **heterochromatin** (highly condensed region). Euchromatin has large amount of DNA material, hence genetically active. Y chromosome has small amount of euchromatin and large amount of heterochromatin, hence it is genetically less active or inert. Both X and Y chromosome show homologous and non-homologous regions. Homologous regions show similar genes while non-homologous regions show dissimilar genes.

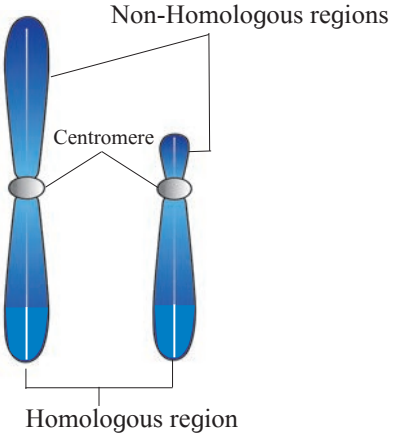


Fig. 3.7 : Structure of X and Y chromosomes (in humans)

Crossing over occurs only between homologous regions of X and Y chromosomes. Non-homologous region of X chromosome is longer and contains more genes than that of

non-homologous region of Y chromosome. X-linked genes are persent on non-homologous region of X-chromosome while Y linked genes are persent on non-homologous region of Y-chromosome.

Can you tell?

1. What are allosomes?
2. Compare X and Y chromosomes.
3. In which region of chromosomes does crossing over take place?

3.8 Linkage and Crossing Over :

Linkage :

It is a known fact that several genes are present on the chromosome. As chromosomes 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 chromosomes that are inherited together is known as **linkage**. Linkage was discovered in plants by Bateson and Punnett and in animals by T. H. Morgan. Linkage is of two kinds - complete and incomplete 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 incompletly 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 chromosome, constitute a linkage group. The number of linkage groups of a particular species corresponds to its haploid number of chromosomes. e.g. *Drosophila melanogaster* has 4 linkage groups that corrrspond to the 4 pairs of chromosomes. Garden pea has 7 linkage groups and 7 pairs of chromosomes.

Sex-linkage :

The transmission (inheritance) of X - linked and Y-linked genes from parents to offspring, is called **sex-linked inheritance**. Sex-linked inheritance is of three types viz. X-linked, Y-linked and XY-linked. Sex linkage is of two kinds :

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. Incomplate 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 blindess, nephritis, retinitis pigmentosa, etc.

Crossing Over :

Crossing over is a process that produces new combinations (recombinations) of genes by interchanging and exhanging 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.

Use your brain power How many linkage groups are in human being and maize?

Know the Scientist :

T. H. Margan (1866-1945)

Thomas Hunt Morgan was an American biologist. He used fruit fly (*Drosophila melanogaster*) in genetic reserch and established the chromosomal theory of heredity. He also discovered the principle of linkage, sex linkage and crossing over.

Margan's work played key role in the field of genetics. He was awarded a Nobel Prize in 1933, in Physiology and Medicine.

- **Morgan's Experiments showing linkage and crossing over :**

Morgan used *Drosophila melanogaster* (fruit fly) for his experiments because, *Drosophila* can easily be cultured in laboratory. It's life span is short, about two weeks. More over, it has high rate of reproduction.

Morgan carried out several dihybrid cross experiments in fruit fly to study genes that are sex-linked. The crosses were similar to dihybrid crosses, as carried out by Mendel in Pea. For example, Morgan and his group crossed yellow-bodied, white eyed female to the wild type with brown-bodied, red eyed males and intercrossed their F_1 progeny.

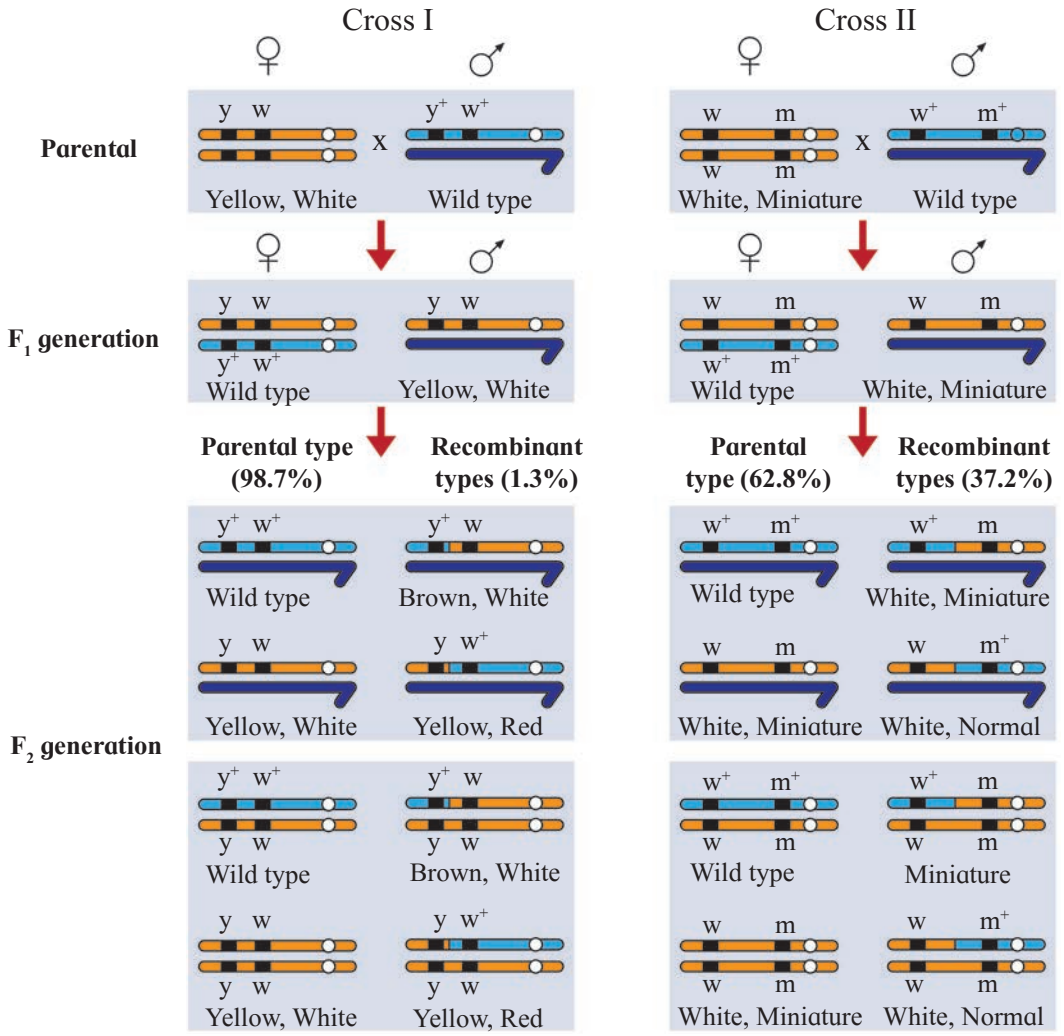


Fig : 3.8 : Linkage and crossing over

He observed that the two genes did not segregate independently of each other and F_2 ratio deviated very signiticantly from 9:3:3:1 ratio.

Morgan and his group knew that the genes were located on X chromosome and stated that when two genes in a dihybrid cross are situated on the same chromosome, then the proportion of parental combination is much higher than non-parental type. This occurs due to physical association or linkage of the two genes. He also found that, when genes are grouped on the same chromosome, some genes are strongly linked. They show very few recombinations (1.3 %). When genes are loosely linked i.e. present far away from each other on chromosome, they show more (higher) recombinations (37.2 %).

For example, the genes for yellow body and white eye were strongly linked and showed only 1.3 percent recombination (in cross-I). White bodied and miniature wings showed 37.2 percent recombination (in cross-II). Cross I shows crossing over between genes y and w. Cross II shows crossing over between genes white (w) and miniature wing (m). Here dominant wild type alleles are represented with (+) sign.

Always Remember

Parental combinations occur more due to linkage and new combinations less due to crossing over.

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 widows 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.

Autosomal recessive traits appear in both sexes with equal frequency. These traits tend to skip generations.

3.10 Sex Linked Inheritance :

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

The inheritance of sex linked genes from parents to their offsprings, is called sex linked inheritance. There are two types of sex-linked genes as X-linked genes and Y-linked genes.

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 charater is transmitted directly from father to son.

Internet my friend

Collect information of Ishihara's Test for colour blindess.

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^cY) 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^CY	X^cY	——
Female	X^CX^C	X^cX^c	X^CX^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.

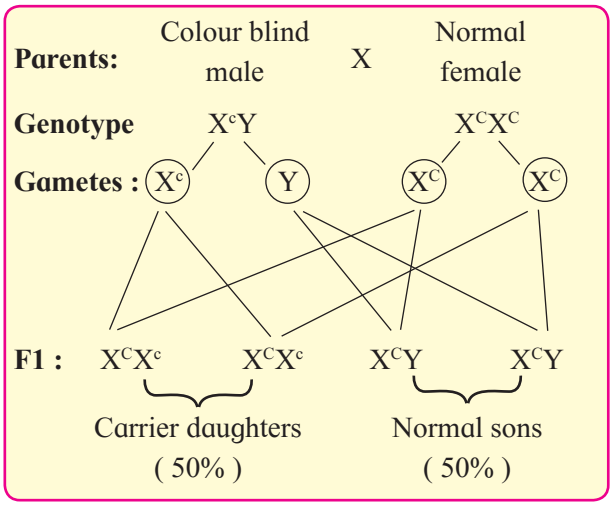


Fig. 3.10 : Sex linked inheritance (colour blindness)

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.

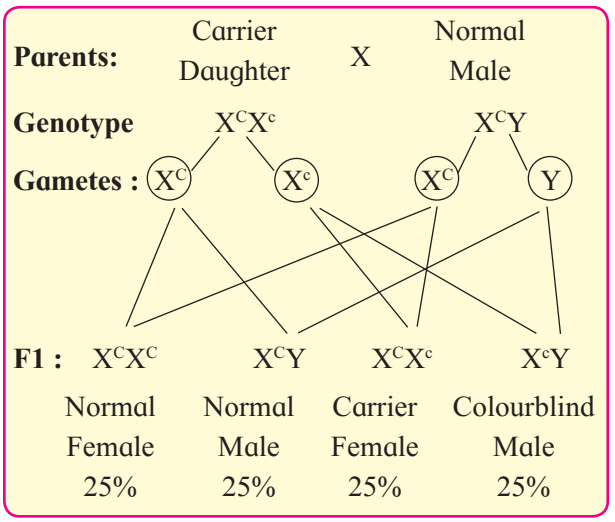


Fig. 3.11 : Sex linked inheritance (colour blindness)

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^HY	X^hY	——
Female	X^HX^H	X^hX^h	X^HX^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.

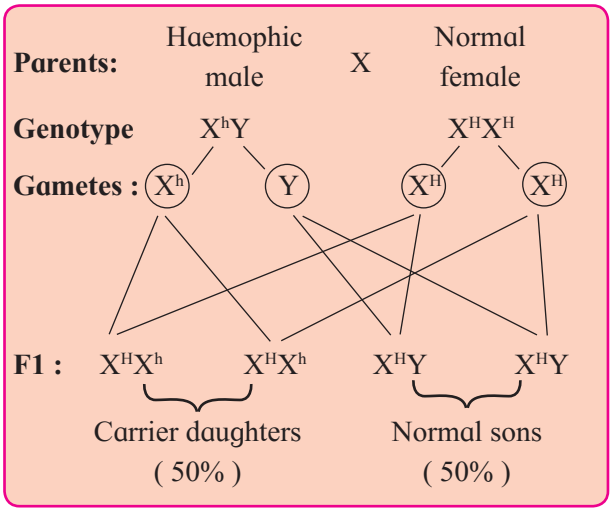


Fig. 3.12 : Sex linked inheritance (Haemophilia)

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

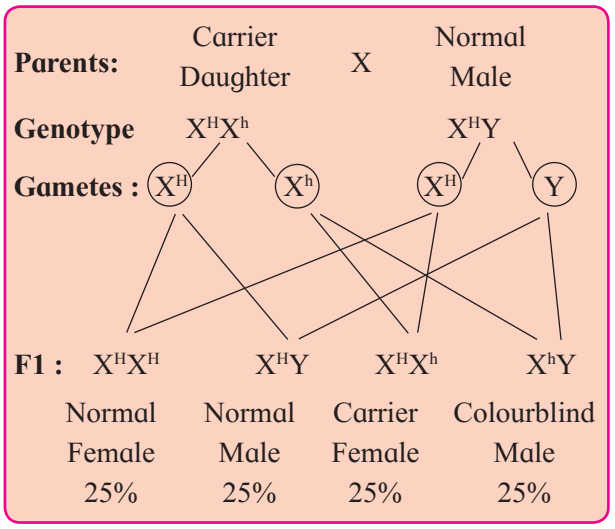


Fig. 3.13 : Sex linked inheritance (Haemophilia)

Do you know ?

Haemophilia is also referred as "The royal disease", because it affected the royal families of England, Germany, Russia and Spain in the 19th and 20th centuries. Queen Victoria of England, who ruled from 1837-1901, was belived to have been the carrier of haemophilia. She passed the trait on to her three of nine children.

Find out

Aarya shows normal blood clotting but her mother is haemophilic. Ramesh shows normal blood clotting but his father is haemophilic. If Ramesh and Aarya were to marry, then find out the posible phenotypes of their offsprings.

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 discribed as **bisexual** or **hermaphrodite** or **monoecious**. 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 infact a chromosome and gave the name 'X-Chromosome'.

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 haplaid 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

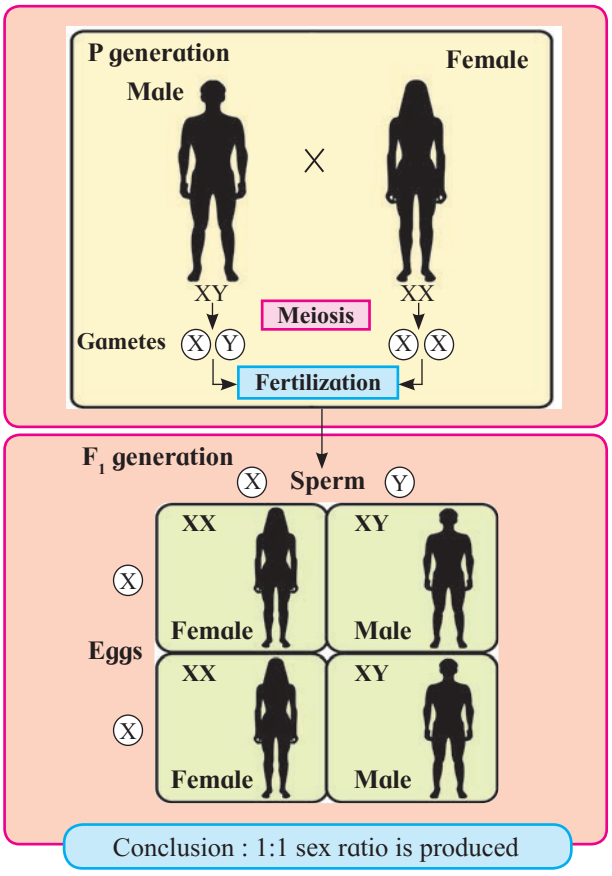


Fig. 3.14 : Sex determination in human beings

sperm containing Y chromosome fertilizes the egg, then diploid zygote is formed that grows into a 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**. Due to lack of knowledge, women are often blamed for giving birth to female child.

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.

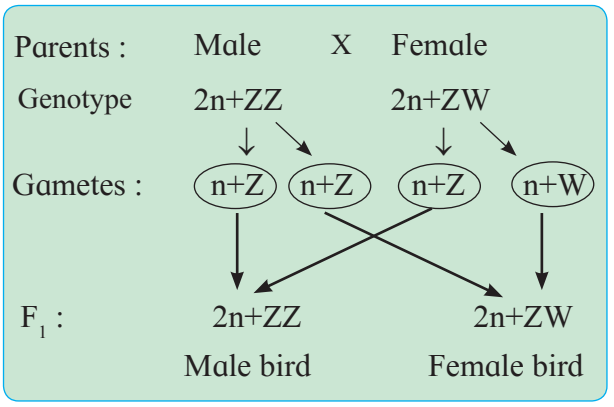


Fig. 3.15 : Sex determination in birds

Something Interesting

In *Bonellia viridis*, the environmental factors determine the sex of individual.
The sex of worm *Bonellia viridis* depends on which location the *Bonellia* larva gets settled.

The marine female *Bonellia worm* has about 10 cm long body. She has a proboscis that can extend over a meter in length. If a *Bonellia* larva settles on the seafloor, it becomes a female.

However when, a larva lands on a female's proboscis and enters the female's mouth, it migrates into her uterus and differentiates into a male. Male lives as parasite in uterus of female fertilizing her eggs.

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 haplaid 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).

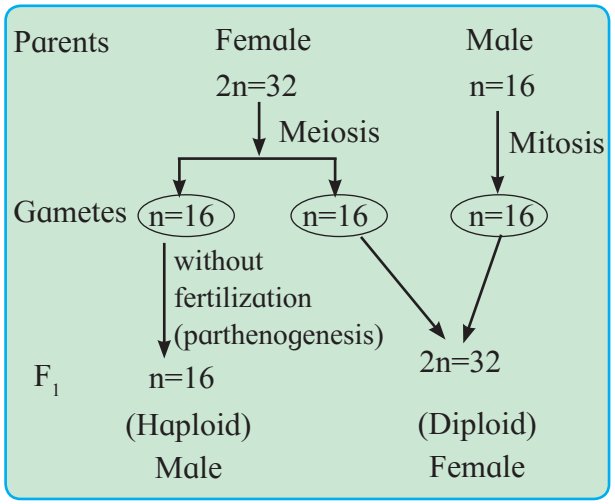


Fig. 3.16 : Sex determination in honey bee

3.12 Genetic Disorders :

Genetic Disorders are broadly grouped into two categories as, **Mendelian disorders** and **chromosomal disorders**, Mendelian discorders 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 arrangment. 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. Massive blood transfusion is needed to these patients. Thalassemia differs from sickle-cell anaemia. The former is a qualitative problem of synthesising few globin molecule, while the latter is a qualitative problem of synthesising an incorrectly functional globin.

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-disjuction 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 palmer crease.

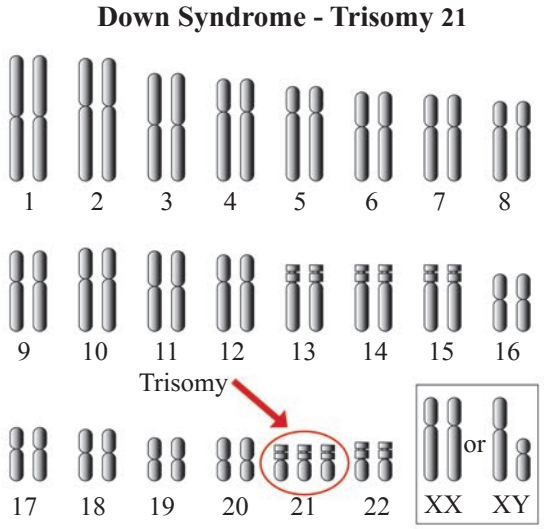


Fig. 3.18 : Karyotype of Down's syndrome

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.

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.

Activity :

Study the complementary and supplementary interaction (digenic interactions) - both in plants and animals

Exercise

Q. 1 Multiple choice questions.

1. Phenotypic ratio of incomplete dominance in *Mirabilis jalapa.*
   a. 2 : 1 : 1 b. 1 : 2 : 1
   c. 3 : 1 d. 2 : 2
2. In dihybrid cross, F_2 generation offsprings show four different phenotypes while the genotypes are .....
   a. six b. nine
   c. eight d. sixteen
3. A cross between an individual with unknown genotype for a trait with recessive plant for that trait is .....
   a. back cross b. reciprocal cross
   c. monohybrid cross d. test cross
4. When phenotypic and genotypic ratios are the same, then it is an example of .....
   a. incomplete dominance
   b. complete dominance
   c. Multiple alleles
   d. cytoplasmic inheritance
5. If the centromere is situated near the end of the chromosome, the chromosome is called .....
   a. Metacentric b. Acrocentric
   c. Sub-Metacentric d. Telocentric
6. Chromosomal theory of inheritance was proposed by .....
   a. Sutton and Boveri
   b. Watson and Crick
   c. Miller and Urey
   d. Oparin and Halden
7. If the genes are located in a chromosome as p-q-r-s-t, which of the following gene pairs will have least probability of being inherited together?
   a. p and q b. r and s
   c. s and t d. p and s
8. Find the mis match pair :-
   a. Down's syndrome = 44 + XY
   b. Turner's syndrome = 44 + XO
   c. Klinefelter syndrome = 44 + XXY
   d. Super female = 44 + XXX
9. A colourblind man marries a woman, who is homozygous for normal colour vision ,the probability of their son being colourblind is –
   a. 0% b. 25%
   c. 50% d. 100%

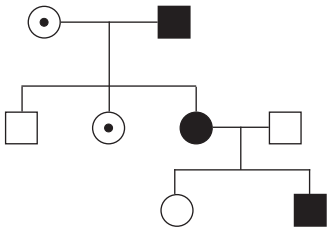
Q. 2 Very Short Answer Questions.

1. Explain the statements :
   a. Test cross is back cross but back cross is not necessarily a test cross.
   b. Law of dominance is not universal.
2. Define the following terms :
   a. Dihybrid cross b. Homozygous
   c. Heterozygous d. Test cross
3. What is allosome?
4. What is crossing over?
5. Give one example of autosomal recessive disorder.
6. What are X-linked genes?
7. What are holandric traits?
8. Give an example of chromosomal disorder caused due to non-disjunction of autosomes.
9. Give one example of complete sex linkage?

Q. 3 Short Answer Questions.

1. Enlist seven traits of pea plant selected/ studied by Mendel.
2. Why law of segregation is also called the law of purity of gametes?
3. Write a note on pleiotropy.
4. What are the reasons of Mendel success?

5. "Father is responsible for determination of sex of child and not the mother". Justify.
6. What is linkage? How many linkage groups do occur in human being?
7. Write note on –PKU
8. Compare - X-chromosome and Y-chromosome.
9. Explain the chromosomal theory of inheritance.
10. Observe the given pedigree chart and answer the following questions.



a. Identify whether the trait is sex linked or autosomal.

b. Give an example of a trait in human beings which shows such a pattern of inheritance.

Q. 4 Match the column-I with column-II and re-write the matching pairs.

Column-I	Column-II
1. 21 trisomy	a. Turner's syndrome
2. X-monosomy	b. Klinefelter's syndrome
3. Holandric traits	c. Down's syndrome
4. Feminized male	d. Hypertrichosis

Q. 5 Long answer type questions.

1. What is dihybrid cross? Explain with suitable example and checker board method.
2. Explain with suitable example an independent assotrment.
3. Define test cross and explain its significance.
4. What is parthenogenesis? Explain the haplo-diploid method of sex determination in Honey bee.
5. In the answer for inheritance of X-linked genes, Madhav had shown carrier male. His answer was marked incorrect. Madhav was wondering why his marks were cut. Explain the reason.
6. With the help of neat labelled diagram, describe the structure of chromosome.
7. What is cris-cross inheritance? Explain with suitable example.
8. Describe the different types of choromosomes.

Project :

Study the genetic traits like Rolling of tongue or Widow's peak in your class and write your own observations.