1. HEREDITY

An elephant resembles other elephants, a rose plant looks like other rose plants, and children resemble their parents, even grandparents or great grandparents. This resemblance among the individuals of a species has given rise to a general truth **'like begets like'** which implies continuity of life. But in human beings, the children often have some individual characters in which they differ from one another, and also from their parents. In fact, their differences are as marked as their resemblances. This is true about other species as well.

The similarities and differences among the members of a species are not coincidental. They are received by the young ones from their parents. The hereditary information is present in the gametes (egg and sperm), which fuse to form the fertilized egg or zygote during sexual reproduction. The zygote then develops into an organism of a particular type. For example, fertilized eggs of sparrows hatch into sparrows only and the fertilized eggs of pigeons hatch into pigeons only. Similarly, a cow gives birth to calves only. The wheat plant gives rise to seeds which in turn, develop into wheat plants. This is the essence of heredity.

HEREDITY

Transmission of characters (resemblances as well as variations) from parents to the offsprings, i.e., from one generation to the next is called heredity.

VARIATION

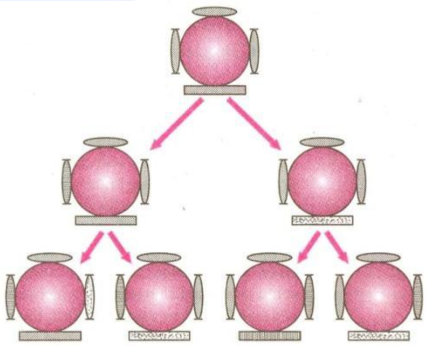
The differences shown by the individuals of a species, and also by the offsprings (siblings) of the same parents are referred to as variations.

1.1 ACCUMULATION OF VARIATION DURING REPRODUCTION

Heredity involves inheritance of basic body design (similarities) as well as subtle changes (variations) in it from one generation to the next generation, i.e., from parents to the offspring. When individuals of this new generation reproduce, the offsprings of second generation will have the same basic body design, the differences that they inherit from first generation as well as newly developed differences.

Asexual reproduction involves single parent. When a single individual reproduces asexually, the resultant two individuals again after sometime reproduce to form four individuals. All these individuals would be exactly similar (carbon copies).

Sexual reproduction, on the other hand, generates even greater diversity. This is so because sexual reproduction involves two parents (father and mother) and every offspring receive some characters of father and some characters of mother. Since, different offsprings receive different combination of characters of their parents (father and mother), they show distinct differences (variations) among themselves as well as from their parents.



Accumulation of variations during reproduction over generations to create diversity

1.2 RULES FOR THE INHERITANCE OF TRAITS

Mendel's Contribution: In human beings, the rules for the inheritance of traits are related to the fact that both the father and the mother contribute practically equal amounts of genetic material to the child. Each trait in the child is influenced by both paternal and maternal DNA (deoxyribo nucleic acid). In other words, for each trait, there will be two versions in each child, one from father and another from mother. Gregor Johann Mendel worked out the basic rules of such inheritance of traits more than a century ago. He studied the inheritance of contrasting characters (traits).

1.2.1 Characters of pea plant studied by Mendel

- Plant height — Tall/short
- Flower colour — Violet/white
- Pod colour— Green/yellow
- Pod shape— Smooth/wrinkled
- Seed colour — Green/yellow
- Seed shape — Round/wrinkled

1.2.2 Mendel's Experimental Plant

Mendel selected garden pea plant (*Pisum sativum*) for series of hybridization experiments because it had the following special features:

(i) It had a short life cycle and, therefore, it was possible to study number of generations quickly.

(ii) Garden pea plant had distinct, easily detectable contrasting variants of features. For instance, some plants were tall and some dwarf; some had violet flowers and some had white flowers; some plants had round seeds and some had wrinkled seeds and so on. Mendel, in fact, noted **seven pairs** of such contrasting characters in garden pea plant.

The characters which always appear in two opposing conditions are called contrasting characters.

(iii) All the contrasting traits existed in every generation because plants had bisexual flowers and normally resorted to self-pollination.

(iv) In these bisexual plants, artificial cross-fertilization could easily be achieved. It was done by removing the stamens (male part) before maturity of the female part of flower and later dusting the pistil (female part) of this flower with the matured pollens from a desired plant.

(v) Each pea plant produced many seeds in one generation.

(vi) The garden pea plants could easily be raised, maintained and handled.

1.2.3 Mendel's Experimental Technique

Mendel conducted breeding experiments in three steps:

(i) Selection of pure parent plants (i.e., plants producing similar traits in every generation).

(ii) Production of first generation of plants by cross breeding (hybridization).

(iii) Raising of second and subsequent generations by self-fertilization of hybrids.

1.2.4 Precautions taken during experiment

While performing his cross breeding experiments, Mendel took a number of precautions. He always focused on the inheritance of the specific traits under consideration and simply ignored others. For instance, in his crosses, only one trait, i.e., size of plant (tall or dwarf) or colour of flower (violet or white) etc. was considered. He called such crosses as **monohybrid crosses.** Mendel also conducted crosses considering two or more contrasting traits of garden pea plant simultaneously. He designated such crosses as **dihybrid crosses, trihybrid crosses** and so on.

In cross breeding experiments, most important precaution required is to avoid self-pollination between two varieties or traits of plants. Mendel removed the anthers (male parts) of the flowers well before the maturity of the female part, i.e., gynoecium of the flowers. This process is called **emasculation.** Such flowers were covered to avoid entry of any foreign pollen grain from outside by wind or animals. For making desired cross, mature pollen grains from the anther of the flower of the desired plant were transferred on the stigma (female part) of the emasculated mature flower. The seeds formed by such crosses were collected. These seeds belonged to the **first filial generation** or **F_1 generation.** To draw effective conclusions, Mendel used the seeds of F_1 generation to raise the F_2 generation by self pollination and also the F_2 seeds for raising F_3 generation by self-pollination. He maintained all the records of his experiments.

1.2.5 Mendel's Monohybrid Cross

A breeding experiment dealing with a single character is called a monohybrid cross.

Mendel first selected **'pure line' plants** (i.e., the plants that produced similar traits generation after generation). He, then, cross-pollinated such plants having the contrasting traits, considering one trait at a time. For instance, in one such cross breeding experiment, he cross bred garden pea plant having round seeds with plant having wrinkled seeds. In this monohybrid cross, the pollen grains from the flower of the desired plant raised from round seeds were transferred over the previously emasculated flower of a plant raised from wrinkled seeds or vice-versa. After the transfer of pollen grains, the cross-pollinated flower was properly covered and seeds produced were allowed to mature. All the seeds of F_1 generation were carefully observed. Mendel observed that all the seeds of F_1 generation were of round type and there were no intermediate characteristics.

He raised plants from F_1 seeds and allowed the flowers to self-pollinate to produce the seeds of F_2 generation. The flowers were kept covered from the beginning to avoid unwanted pollens to reach these flowers. In F_2 generation, Mendel observed the appearance of both round and wrinkled seeds in approximately 3 : 1 ratio.

Parental Generation — Male pea plant with round seeds × Female pea plant with wrinkled seeds

↓ (Cross pollination)

F_1 Generation — All plants with round seeds

↓ (Self pollination)

F_2 Generation — Round Seeds : Wrinkled Seeds

3 : 1

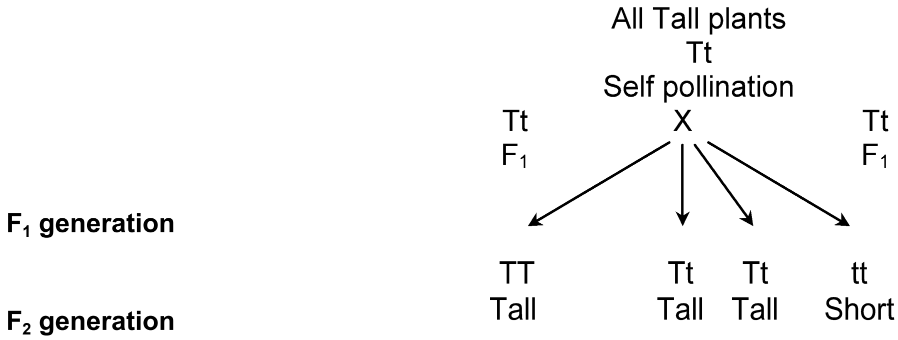
A similar ratio was seen when he took the character for tallness or shortness of a pea plant.

Monohybrid cross : Cross between 2 pea plants with one pair of contrasting characters Tall/short (monohybrid cross)

Tall plant X Short plant

TT ↓ tt

Parents

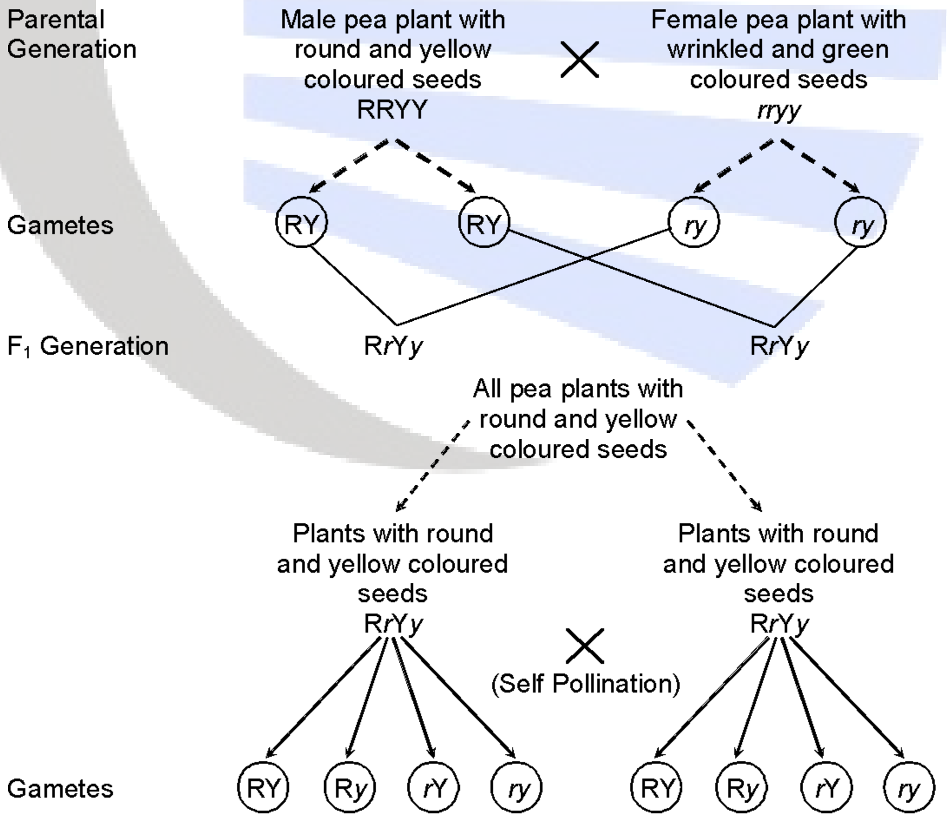


1.2.6 Mendel's Dihybrid Cross

Mendel also studied the inheritance of two characters simultaneously.

A breeding experiment dealing with two characters at the same time is called a dihybrid cross.

In one such cross, Mendel considered shape as well as colour of the seeds simultaneously. He selected pure line plants and then cross-pollinated flowers raised from seeds of round shape and yellow colour with those from wrinkled seeds and green colour. Mendel observed that in F_1 generation all seeds had the features of only one parental type, i.e., round shape and yellow colour. He raised plants from F_1 generation seeds and allowed the flowers to self, pollinate to produce the seeds of F_2 generation. These flowers were kept covered from the beginning. In F_2 generation, Mendel observed the appearance of four types of combinations. These included **two parental types** (round shaped yellow coloured seeds and wrinkled shaped and green coloured seeds) and **two new combinations** (round shaped green coloured seeds, and wrinkled yellow coloured seeds) in 9 : 3 : 3 : 1 ratio. The dihybrid cross is as follows :



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

F_2 Generation	Plants with round and yellow coloured seeds		Plants with round and green coloured seeds		Plants with wrinkled and yellow coloured seeds		Plants with wrinkled and green coloured seeds
	9	:	3	:	3	:	1

1.2.7 Mendel's Conclusions

Based on the findings of monohybrid and dihybrid crosses, Mendel concluded that –

(i) In a **monohybrid cross,** only one of two contrasting characters (traits) appeared in F_1 generation. However, in F_2 generation, both the parental traits appeared in certain proportion (3:1).

(ii) **In a dihybrid cross,** when two contrasting pairs of traits were considered simultaneously, only one parental combination appeared in F_1 generation. However, in F_2 generation, raised by self-pollination, four combinations of traits appeared. These included two parental type traits and two new combinations in approximately same proportion.

1.2.8 Mendel's Interpretation

On the basis of monohybrid and dihybrid crosses, Mendel postulated that :

(i) There is a pair of unit **factors** controlling each character in pea plant, one inherited from each parent. Mendel considered these factors as the carriers of hereditary information from one generation to another, i.e., from parents to the offsprings. At the time of reproduction, when gametes are formed, these factors segregate so that each gamete receives only one factors of each character. This is called **law of segregation.** Fertilization brings these two factors again together in the offspring.

(ii) In F_1 generation, only one character was expressed. Mendel called it as **dominant** character. The character which was not expressed was termed **recessive** character. **This phenomenon of appearance of only one of two contrasting traits in F_1 generation is termed as the law of dominance.**

(iii) The characters are not lost even when they are not expressed.

(iv) When F_1 offsprings were allowed to be self pollinated, both the parental traits were expressed in definite proportion in F_2 generation.

(v) From the F_2 generation of a dihybrid cross, Mendel postulated that inheritance of factors controlling a particular trait in an organism is independent of the other. **This is called law of**

independent assortment.

Hence, at the time of reproduction, two pairs of factors of each of the two traits in a dihybrid cross segregated independently during gamete formation and randomly formed combinations in the F_2 generation. This is why new combinations appeared along with parental combinations.

2. STRUCTURE OF DNA (DEOXYRIBO NUCLEIC ACID)

The expanded form of DNA is deoxyribonucleic acid. It was first isolated by the scientist Frederick Meisher from the nucleus of the pus cells in 1869. He named it as 'nuclein' or nucleic acid because of its acidic nature. Later, it was experimentally proved by the scientists Griffith (1928), Avery, McLeod and McCarty (1944) that DNA is the carrier of the genetic information from generation to generation. It transmits the hereditary characters in a coded language from parents to the offsprings (i.e., from one generation to another).

DNA is a macromolecule or polymer. It is made of very large number of 'nucleotide' units and hence is termed polynucleotide.

Each nucleotide unit in a DNA molecule is made up of three components:

1. **Deoxyribose sugar:** It is a pentose sugar represented as S.
2. **Nitrogenous base:** Each nucleotide unit has a nitrogen-containing base. In a DNA molecule, nitrogenous bases are of two types.
   (a) ***Purines***: The purines in a DNA molecule are – **Adenine** (A) and **Guanine** (G).
   (b) ***Pyrimidines***: The pyrimidines in a DNA molecule are – **Cytosine** (C) and **Thymine** (T)
3. **Phosphate group:** The phosphate group contains one phosphorus atom and four specifically linked oxygen atoms. It is represented as P.

Thus, there are four types of nucleotides in a DNA molecule depending upon the kind of nitrogenous base present in each nucleotide.

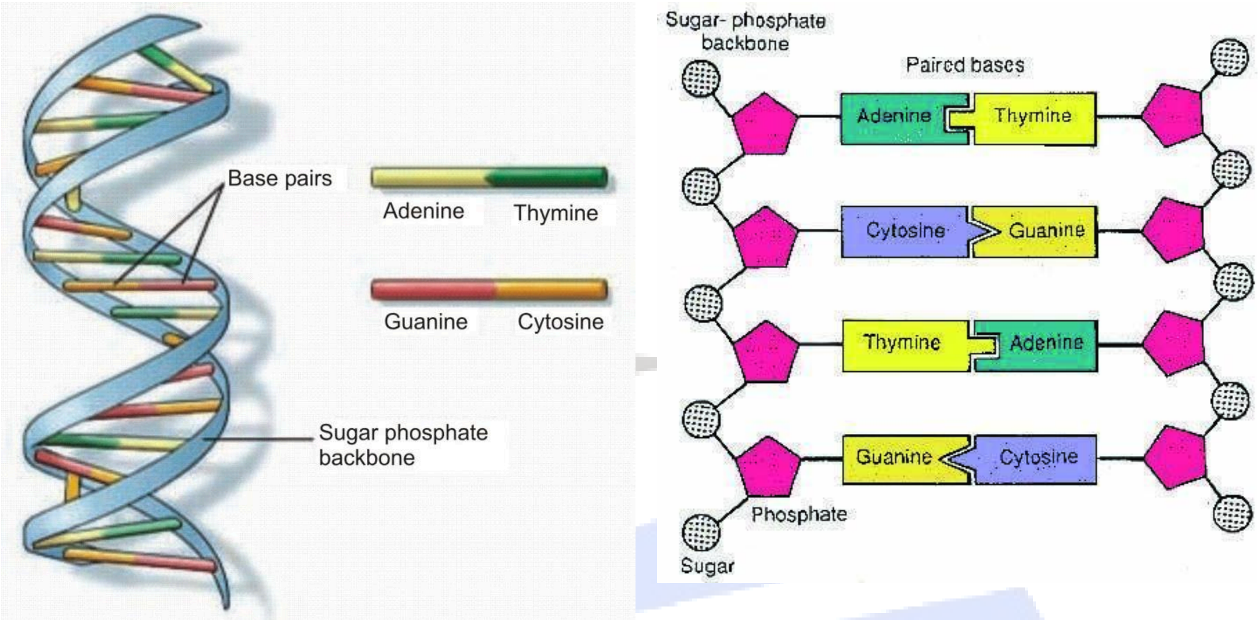
2.1 DOUBLE HELICAL MODEL OF DNA

J.D. Watson and F.H.C. Crick proposed **the double helical model of DNA** in 1953. They were awarded the Nobel Prize for this discovery in 1962. The important features of the double helical model are:

(i) DNA molecule is made up of **two long polynucleotide strands** forming a **double helical structure (double helix)** just like a spiral staircase. Each helical turn of the DNA molecule is **3.4 nm** in length in which **ten nucleotide** base pairs are present.

(ii) **Deoxyribose sugar** and **phosphate molecules** are joined alternately to form the backbone of each polynucleotide strand. The **nitrogenous base** of each nucleotide is attached to the sugar molecule and projected towards the interior of the double helix.

(iii) In the interior of double helix, the nitrogenous bases of two polynucleotide strands form a pair with the help of hydrogen bonds. **Adenine (A)** always pairs with **thymine (T)** and **guanine (G)** always pairs with **cytosine (C).**



(a) (b)

Structure of DNA

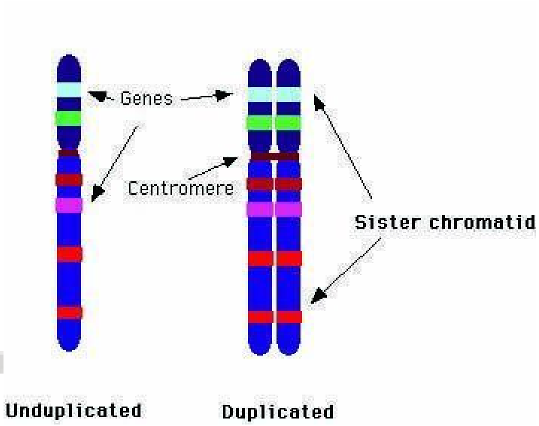
Thus, the two polynucleotide strands of the DNA molecule are joined by hydrogen bonds between specific nitrogenous bases. **Such a specific pairing of the bases of the opposite strands of the DNA molecule is called complementary pairing. Adenine (A) and thymine (T) are complementary to each other. Similarly, guanine (G) and cytosine (C) are complementary to each other.** The hydrogen bonding between the specific nitrogenous bases keeps the two strands to hold together. Therefore, all the base pairs remain stacked between the two strands.

2.2 GENES

Mendel was the first scientist to visualise a gene as a unit of inheritance in 1866. He called it **factor.** The word **'gene'** was, however, introduced by Johannsen in 1909.

Earlier knowledge about genes was based on the studies conducted on fruit fly (*Drosophila melanogaster*) and some higher plants. It was then considered as **the unit of inheritance.** However, nothing was known about its structure. Once the molecular structure of chromosomes and DNA was established, our knowledge about genes also increased. It is now well known that genes are nothing but **segments of DNA on a chromosome occupying specific positions.** Approximately 30,000 – 40,000 genes are present on 46 human chromosomes.

Chemically, each gene has a specific sequence of nucleotides, which determines its functional property. It is the genes which determine everything about us, our complexion, the shape and colour of our eyes, the shape of our nose and chin etc. Actually, gene is a segment of DNA molecule, which has coded information to form a particular protein in the cell. This protein can function either as a structural protein or as a functional protein (enzyme).



Location of genes on a chromosome

2.3 MECHANISM OF HEREDITY

Genes control characters or traits of an organism. Lets understand the mechanism of heredity:

A section of **cellular DNA**
↓
Gene
↓
Provides information for Synthesis **of protein**
↓
Protein controls a character

For example:

Gene T $\xrightarrow{\text{provides information}}$ Synthesis of **efficient enzymes** (protein)
↓
Tall plant ← **More production** of growth hormone

Gene t $\xrightarrow{\text{provides information}}$ Synthesis of **less efficient** enzyme
↓
Short plant ← **Less production** of growth hormone

On the basis of modern molecular concept, a gene is a hereditary unit, a segment of DNA having specific sequence of nucleotides that determines its specific biological function. It can maintain constancy from generation to generation but at times may also undergo sudden mutations (inheritable changes) to bring variations.

2.4 INHERITANCE OF GENES FROM PARENT PLANTS TO OFFSPRINGS

New mixed characters in progeny in F_2 generation in Mendel's experiment showed that **some genes are linked and always inherited together.** This showed that **genes are located on chromosomes.** One set of chromosomes (with one set of genes) known as germ cell is contributed by each parent to the child.

Parents **Male**
2 sets of genes
(on 2 sets of chromosomes = 23 pairs)
↓ Meiosis
Male germ cell
(One set of genes on
one set of chromosome)

Female
2 sets of genes
(on 2 sets of chromosomes =23 pairs)
↓ Meiosis
Female germ cell
(One set of gene on
one set of chromosome)

Fertilization
↓
Child
2 sets of genes on 2 sets of chromosomes (23 pairs)
(one paternal set of genes + one maternal set of genes)
(23 chromosomes + 23 chromosomes)

3. SEX DETERMINATION

In sexual reproduction, **male** and **female gametes** fuse during fertilization to form **zygote.** This zygote then develops into the offspring (male or female). The question that comes to our mind is as to how the sex of the offspring is determined in the zygote? We now know, that genetics is involved in the determination of the sex of an individual.

The mechanism, by which the sex of an individual is determined as it begins life, is called sex determination.

In diploid organisms having separate sexes, a specific pair of chromosomes in each diploid cell, determines the sex of the individual. They are called **sex chromosomes.** All other chromosomes are termed **autosomes** as these have genes, which control the somatic (body) characters. The two members of each pair of **homologous autosomes** are similar in size and shape, but this may not be true with sex chromosomes. For instance, in human beings, there are 46 chromosomes. Of these, 44 (22 pairs) are autosomes and 2 (one pair) are sex chromosomes. The sex chromosomes in human beings and also in fruit-fly (*Drosophila*), are of two types – **X chromosome** and **Y chromosome.**

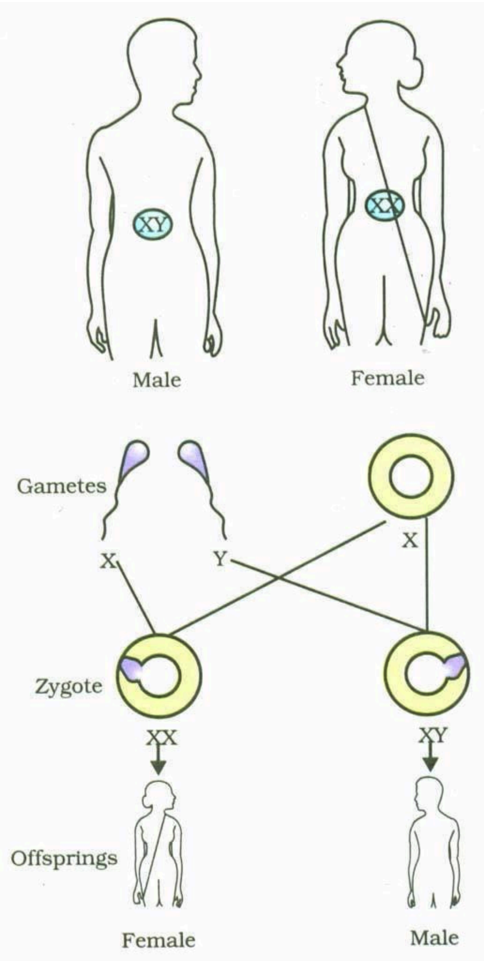
(i) A male individual contains one X chromosome and one Y chromosome i.e. XY (heteromorphic sex chromosomes having different size and shape). Male produces two different kinds of gametes (sperms); half of the gametes having X chromosome and other half having Y chromosomes. Therefore, male is called heterogametic.

(ii) A female individual contains two similar X chromosomes i.e., XX (homomorphic sex chromosomes of same size and shape). Female, therefore, produces same type of all gametes (ova or eggs). So, female is called homogametic.

The sex of the child is determined at the time of fertilization when male and female gametes fuse to form zygote.

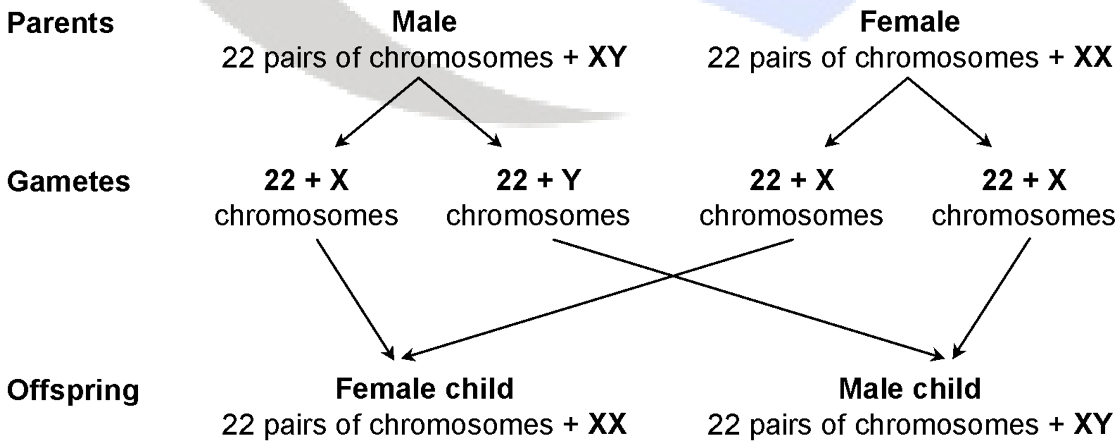
If a sperm (male gamete) carrying X chromosome fertilizes an egg or ovum (female gamete) carrying X chromosome, then the offspring will be a girl (female). This is because the offspring will have XX combination of sex chromosomes.

If a sperm (male gamete) carrying Y chromosome fertilize an egg or ovum (female gamete) which has X chromosome, then the offspring will be a boy (male). This is because, the offspring will have XY combination of sex chromosomes.



Sex determination in human beings

So, we can conclude that the sex of the child (offspring) is determined by the type of sperm that fuses with ovum at the time of fertilization. Therefore, there is 50% chance of a male child being born and a 50% chance of a female child being born.



3.1 ROLE OF ENVIRONMENTAL FACTORS IN SEX DETERMINATION

Sex determination is sometimes regulated by environmental factors also. In some reptiles, the temperature at which the fertilized egg is incubated before hatching is important environmental factor for determining the sex of the offspring. Two examples are cited below:

(i) **First example** is of a turtle (*Chrysema picta*). In this species, high incubation temperature results in development of female progeny.

(ii) **Second example** is of a lizard (*Agama agama*). In this species, high incubation temperature results in male progeny.

In other animals, e.g., snails, individuals can change sex, indicating that sex is not genetically determined in this case. In plants, genetic basis of sex determination exists but it is different from that of animals.