Genetics - Some Basic Fundamentals

Genetics

It is the study of transmission of characters from parents to offspring and the laws relating to such transmission.

Heredity

The phenomenon of passing of characters from parents to progeny through successive generations is called **heredity**.

Variation

The difference in the characters or traits among the individuals of a species is called **variation**.
For example, in most people, the earlobe is hanging and is called a free earlobe. However, in some people, the earlobe is closely attached to the side of the head, and it is called an attached earlobe.

Free Earlobe

Attached Earlobe

Chromosomes

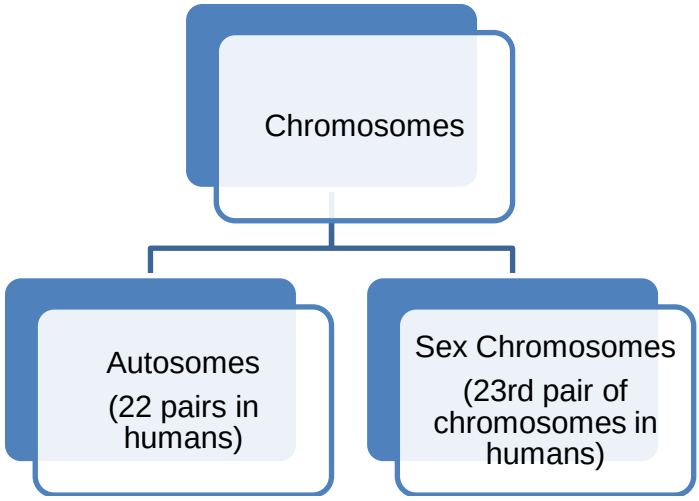
The chromosome is a thread-like structure in the nucleus of a cell. It is formed of DNA and carries the genes.

The chromosome number is constant for the individuals of a species. For example, humans have 46 chromosomes.

Some more examples are given in the following table.

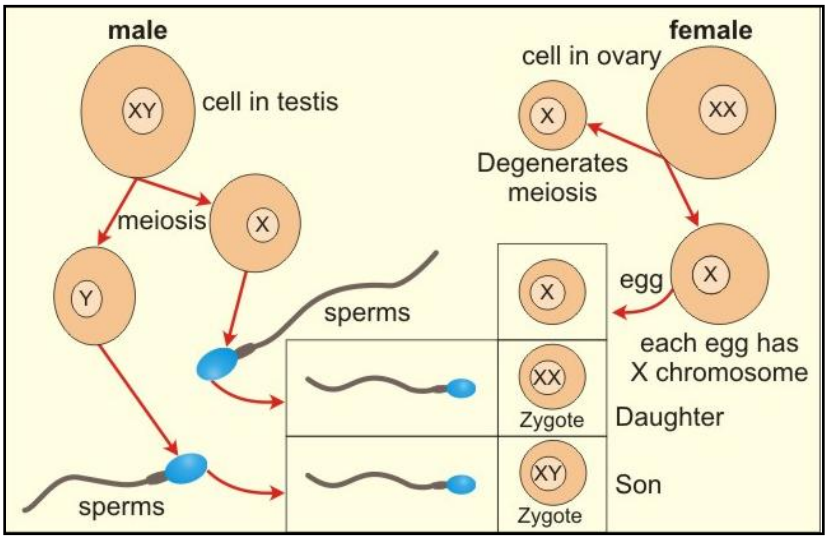
Organism	Number of Chromosomes
Mouse	40
Gorilla	48
Monkey	54
Dog	78
Onion	16
Maize	20

A pair of corresponding chromosomes of the same shape and size, one from each parent, is known as **homologous chromosomes**.



Sex Determination

- The process by which the sex of a person is determined is called **sex determination**.
- There are two types of sex chromosomes—X chromosome and Y chromosome.
- A male has one X chromosome and one Y chromosome (XY).
- A female has both X chromosomes (XX).



If a sperm carrying the X chromosome fertilises an ovum which always carries the X chromosome, then the combination of sex chromosomes will be XX, and hence, the child born will be a female (girl).

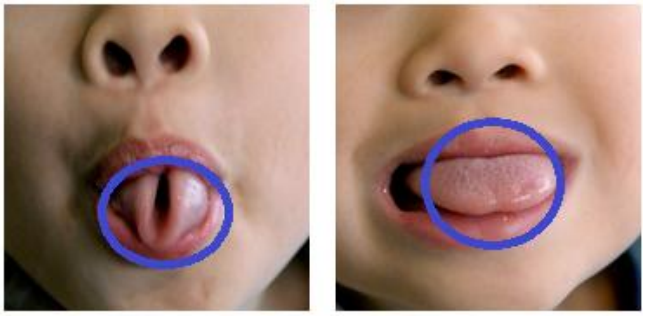
If a sperm carrying the Y chromosome fertilises an ovum, then the combination of sex chromosomes will be XY, and hence, the child born will be a male (boy).

Thus, the male (father) is responsible for the sex of the baby.

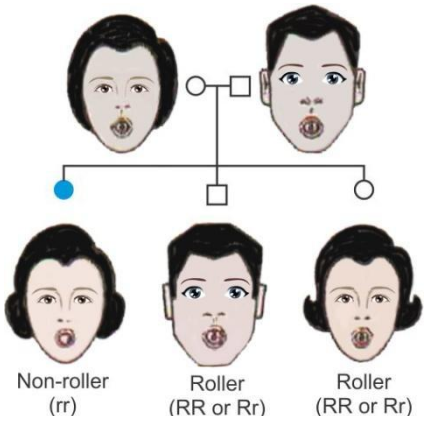
Genes

- Genes are specific DNA segments on a chromosome which determine hereditary characteristics.
- They are responsible for the transfer of characteristics from parents to their offspring.
- Genes work in pairs.
- Every gene has two alternative forms for a character producing different effects; these alternative forms are called **alleles**.
- The alternative forms of a character are known as **traits**.
- The gene which decides the appearance of an organism even in the presence of an alternative gene is known as a **dominant gene**.
- The gene which decides the appearance of an organism only in the presence of another identical gene is called a **recessive gene**.
- The combination of genes present in an organism is called a **genotype**.
- Characteristics which are genetically controlled and are visible in an organism are called a **phenotype** of an organism.
- In the character of tongue rolling, there are two possibilities—one can either roll or cannot roll the tongue.

An Example of Inheritance – Tongue Rolling



- The gene for tongue rolling which is dominant is 'R', and the gene for non-rolling which is recessive is 'r'.
- Thus, for the phenotype tongue rolling, there are two kinds of genotype—homozygous dominant, a pair of similar alleles (RR), and heterozygous dominant, a pair of dissimilar alleles (Rr).



Symbols: ○ – Female; □ – Male

Hollow symbols represent the usual expressed characters. Here, it is the rolling of the tongue.
Solid symbols represent the unusual expressed characters. Here, it is the non-rolling of the tongue.

In the above chart, both father and mother are tongue rollers. Of their three children, two are tongue rollers and one cannot roll the tongue.

The recessive allele ‘rr’ in the third child for non-rolling indicates that in ‘rr’ the child must have received one ‘r’ from each parent. Therefore, each parent is heterozygous for tongue rolling (Rr).

Sex-linked Inheritance

Sex-linked inheritance is the appearance of a trait which is due to the presence of an allele exclusively either on X chromosomes or on the Y chromosome.

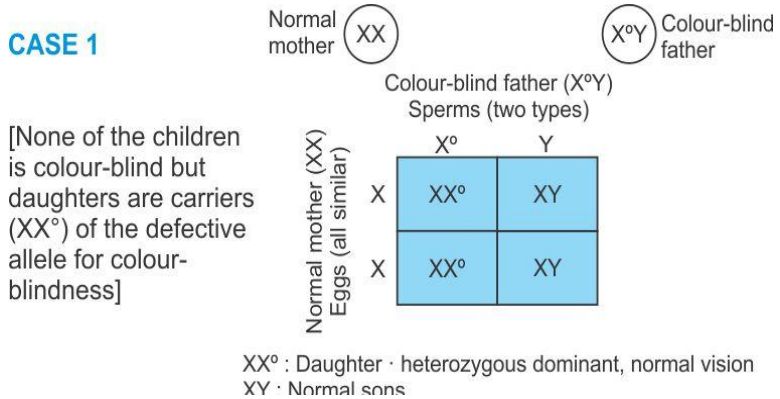
X-linked Inheritance

Defects occur due to recessive genes, which are on the X chromosome.
For example, colour blindness and haemophilia.

- **Haemophilia**
  In haemophilia, the patient is at a risk of bleeding to death because the blood either fails to clot or takes time to clot.

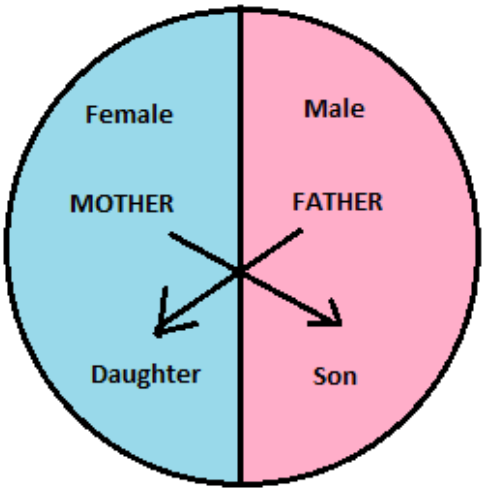
- **Colour blindness**
  A colour blind person fails to distinguish red and green colours.

  Case 1: Normal mother (XX) × Colour blind father (X^oY)



Criss-cross Inheritance

X-linked inheritance is also called **criss-cross inheritance**.
This is because the son may inherit a trait from the normal carrier mother.



Y-linked Inheritance

Traits occur due to dominant genes which are on the Y chromosome. Such traits occur only in males. For example, hypertrichosis (hair growth on ears) and baldness.

Hypertrichosis

Baldness

Mendel's Experiments on Inheritance

Gregor Mendel systematically studied the patterns of inheritance which involved the transfer of characters from one generation to the next. He is known as the **Father of Genetics**.

Gregor Mendel

Mendel conducted breeding experiments on *Pisum sativum* (garden pea).
He selected the pea plant for three reasons:

- Many varieties available in alternative forms of a character.
- Pure forms which bred true, i.e. produced the same type generation after generation.
- Garden pea is self-pollinating but could be cross-pollinated artificially.

Following are the seven pairs of contrasting characters in the garden pea studies by Gregor Mendel.

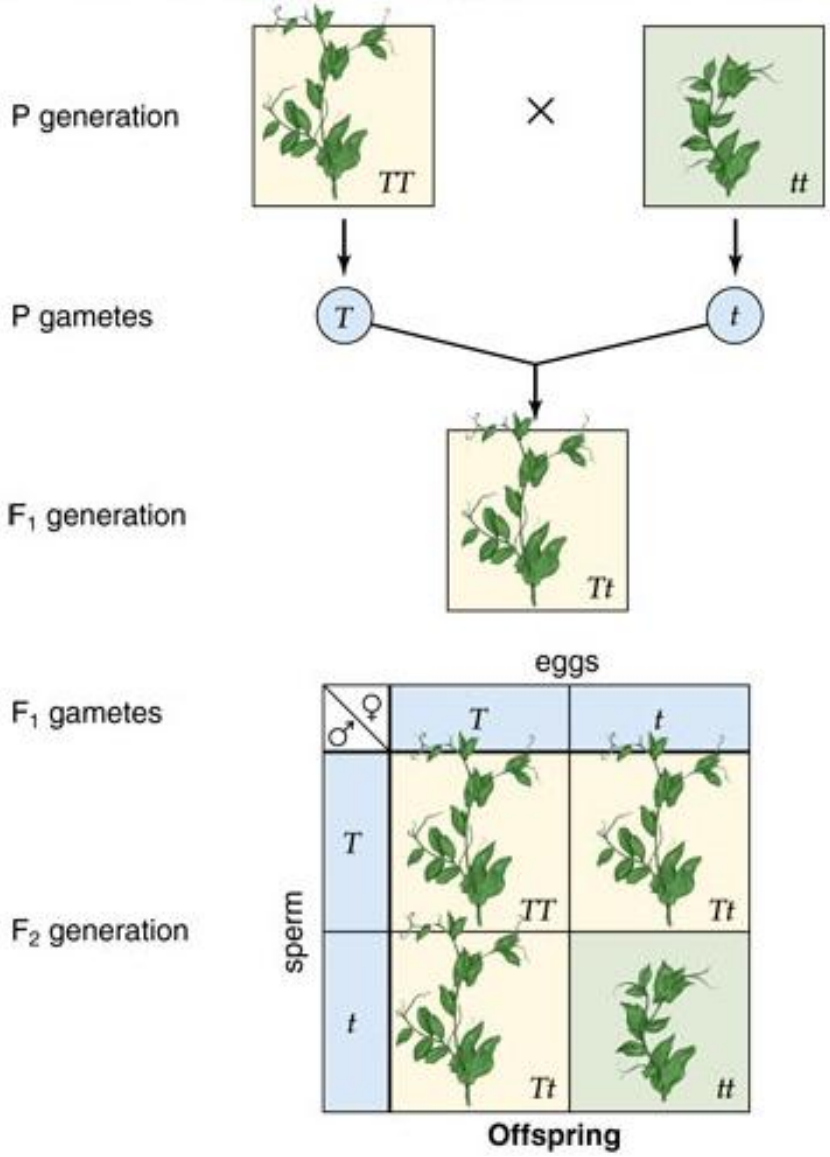
	Height	Seed Shape	Seed Color	Seed Coat Color	Pod Shape	Pod Color	Flower Position
Dominant	Tall	Round	Yellow	Green	Inflated (full)	Green	Axial
Recessive Trait	Short	Wrinkled	Green	White	Constricted (flat)	Yellow	Terminal

Mendel's findings are now called as **Mendel's Laws of Inheritance**.

Monohybrid Cross

Mendel first carried out cross breeding by taking only one character.

Example: Pure tall (TT) pea plant was crossed with **dwarf (tt)** plant.



Phenotypic Ratio: 3:1

Genotypic Ratio: 1:2:1

- F_1 Generation: All plants obtained were heterozygous tall. Plants from the F_1 generation were self-pollinated to obtain the F_2 generation.
- F_2 Generation: One homozygous tall plant, two heterozygous tall plants and one homozygous dwarf plant were obtained.

The ratio obtained by crossing for two different traits of a single character is known as **monohybrid ratio**.

Dihybrid Cross

Mendel carried out the cross breeding by taking two characters together.

Example: Plant with **round yellow seeds (RRYY)** was crossed with the plant bearing **wrinkled green seeds (rryy).**

- F_1 Generation: All plants obtained were heterozygous tall. Plants from the F_1 generation were self-pollinated to obtain the F_2 generation.
- F_2 Generation: One homozygous tall plant, two heterozygous tall plants and one homozygous dwarf plant were obtained.
- The phenotypic ratio was 9:3:3:1.

The ratio obtained by breeding two pairs of contrasting characters is called **dihybrid ratio**.

Mendel's Laws of Inheritance

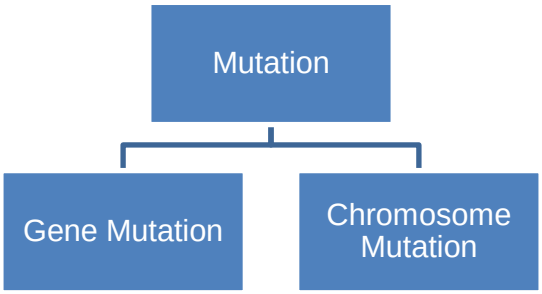
Law of Dominance	Out of a pair of contrasting characters present together, only one is able to express itself while the other remains suppressed.
Law of Segregation	Two members of a pair of factors separate during the formation of gametes.
Law of Independent Assortment	In case of two pairs of contrasting characters, the distribution of the members of one pair into the gametes is independent of the distribution of the other pair.

Application of Mendel's Laws

- Gives an idea of new combination in the new generation.
- Enables to predict the frequency of characters in the new generation.
- Better quality plant and animal breeds can be produced.
- By the technique of hybridisation, new type of plants with new combination of useful characters can be produced.

Mutation

Mutation is the sudden change in one or more genes, or in the number or in the structure of chromosomes.



Sickle Cell Anaemia

It a blood disease caused due to gene mutation. The mutation causes changes in DNA and results in the production of sickle-shaped RBCs.

Down's Syndrome

It is a mental deficiency caused due to one extra chromosome (chromosome no. 21).

Radioactive Radiations

Radioactive radiations alter the gene structure. The atomic explosions which occurred during World War II in 1945 in Japan led to a number of deformities in the body of animals and plants.

Modern Applications of Genetics

- Genetic engineering is the technique in which the genetic constitution of an organism is altered by introducing new genes in its genome.
- Genetically modified organisms are obtained by genetic engineering. Hormone insulin is the first product produced by using genetic engineering.