Genetics

Genetics is the study of variations and how they are transferred from one generation to another.

Gregor Johann Mendel is considered to be the **father of genetics**. During his time, his findings were not accepted, but later in 1900, three scientists **DeVries, Correns,** and **Tschermak** rediscovered Mendel's work.

The term genetics was coined by W. Batson in 1905.

Now let us explore some of the terms related to the study of genetics.

- **Heredity** − It is the transmission of traits from one generation to the other generation.
- **Variation** − It can be defined as the difference observed among members of the same species and also among offsprings of the same parent.
- **Gene** − A gene is the unit of inheritance, which is transferred from the parent to the offspring. It controls the expression of a character. A gene is a linear piece of DNA which is present in nucleus.
- **Allelomorphs** − Every character is controlled by two genes, which control contrasting expressions. A pair of genes that controls the contrasting characters and lies on the same loci in the homologous chromosomes is called an allele.
- **Dominant allele** − An allele which expresses itself in the presence of its contrasting allele is called a dominant allele. For example, the character tallness is determined by two alleles **T** and **t**, where **T** is the dominant allele and **t** is the recessive allele. In the presence of T, the expression of t does not occur.
- **Recessive allele** − The allele which cannot express itself in the presence of the dominant allele is called a recessive allele.
- **Homozygous organism** − In an individual, if the alleles of a character are similar, then they are known as homozygous. For example, **TT** is a homozygous condition.
- **Heterozygous organisms** − If the alleles of a character are dissimilar, then they are called heterozygous. For example, **Tt** is a heterozygous condition.
- **Phenotype** − It is the physical expression of a character. E.g., tall plant
- **Genotype** − It represents how an organism is genetically made up. For e.g., **TT** or **Tt** or **tt**

- **F_1 generation** − It is the first filial generation which is produced when two pure parents are crossed. For example, the F_1generation produced when two pure line plants **TT** and **tt** are crossed is **Tt.**

- **F_2 generation** − It is the second filial generation of progeny formed when two F_1 generation plants are crossed.

Mendel and His Experiments

The individuals of a family (parents and offspring) have more similarity in comparison to others. This is because certain characteristics are passed from the parents to the off springs without any variation.

Heredity is defined as the transmission of characteristics from one generation to another. These characteristics may be physical, mental, or physiological.

Commonly observed heritable features are curly hair, a particular type of ear lobe, hair on ears etc.

Transmission of traits from the parents to progeny - Mendel's Work

Gregor Johann Mendel (1822 – 1884) was the first to carry out the study on the transmission of characteristics from the parents to the offsprings. He proposed that heredity is controlled by factors, which are now believed to be segments of chromosomes or genes.

Mendel performed experiments on a garden pea (*Pisum sativum*) with different visible contrasting characters. He selected seven contrasting pairs of characters or traits in a garden pea.

These include round/wrinkled seeds, tall/short plants, green/yellow pod colour, purple/white flower colour, axial/terminal flower, green/yellow seed colour, and inflated/pinched ripe pods.

Mendel's experiment

Mendel performed experiments in three stages:

Mendel's experiment
- Selection of true breeding parents
- Obtaining F_1 plants
- Self pollination of F_1 plants to generate F_2 and F_3 plants

Selection of parents: Mendel selected true breeding pea plants with contrasting characteristics for his experiment.

True breeding plant is the one that produces an offspring with the same characteristics on self-

pollination. For example, a tall plant is said to be true breeding when all its progeny formed after self-pollination are tall.

Production of F_1 plants: F_1 generation is the first filial generation. It is formed after crossing the desirable parents. For example, Mendel crossed a pure tall pea plant with a pure dwarf pea plant. All F_1 plants were found to be tall.

Results of self-pollination of F_1 plants: Mendel found that on self-pollination of F_1 plants, the progenies obtained in F_2 generations were not all tall plants. Instead, one-fourth of F_2 plants were found to be short.

Mendel's explanation for the reappearance of the short trait:

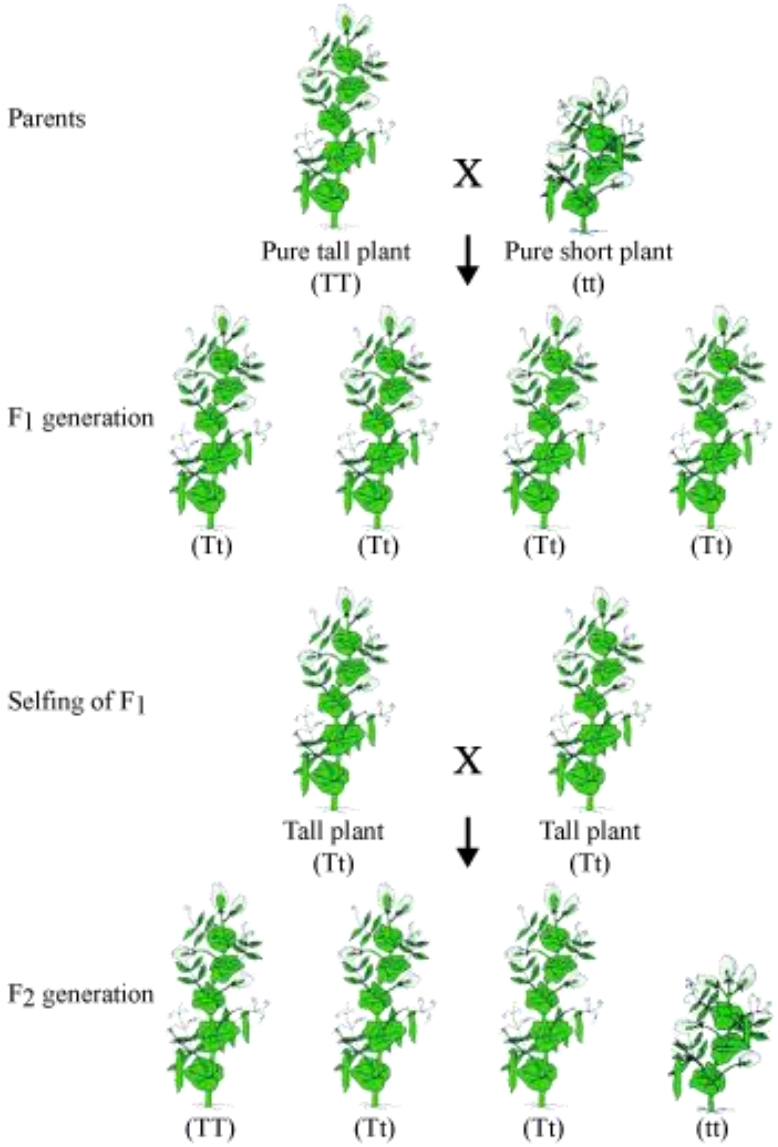
From this experiment, Mendel concluded that F_1 tall plants were not true breeding. They were carrying both short and tall height traits. They appeared tall, because tall trait was dominant over short trait.

Dominant trait: It is a trait or characteristic, which is able to express itself over another contrasting trait. For example, tall plants are dominant over short plants.

Recessive trait: It is a trait which is unable to express its effect in the presence of the dominant trait.

Mendel represented the dominant trait as upper case **T** (i.e. T for tallness), and the recessive trait as lower case **t** (i.e. t for shortness). These traits are actually the genes present in the chromosomes of a cell.

Thus, Mendel's experiment can be represented as follows:



Revival of the trait that was unexpressed in F_1 (dwarf) was observed in some F_2 progeny. Both traits, tall and dwarf, were expressed in F_2 generation in ratio 3:1.

Mendel proposed that something is being passed unchanged from generation to generation. He called these things as ‘factors’ (presently called genes). Factors contain and carry hereditary information.

Traits may not show up in an individual but are passed on to the next generation.

Inheritance of traits over two generations

The appearance of F_1 plants was similar to their parents i.e. they were tall, but were actually different from their parents. Mendel introduced the terms **genotype** and **phenotype**.

Genotype is the genetic constitution of an organism, which includes all genes that are inherited from both the parents. For example TT, Tt, and tt are genotypes of organisms with reference to their height.

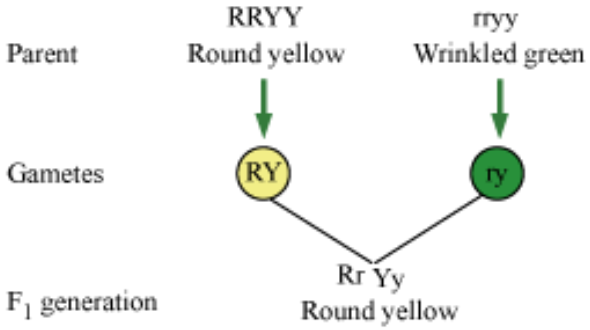
Phenotype is the observable trait or characteristic of an organism, which is the result of genotype. For example, tallness and shortness are phenotypes resulting from different genotypes.

The above experiment of Mendel involved only one pair of contrasting characters (tall/short plant height), so it is called a **monohybrid cross.**

If two pairs of contrasting characters are involved, then the cross is termed as **dihybrid cross**

Inheritance of Two Genes (Dihybrid Cross)

- In dihybrid cross, we consider two characters. (e.g., seed colour and seed shape)
- Yellow colour and round shape is dominant over green colour and wrinkled shape.



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

Phenotypic ratio − 9:3:3:1

Round yellow − 9

Round green − 3

Wrinkled yellow − 3

Wrinkled green −1

Mendel's Laws of Inheritance

Principles of Mendel:

- Each characteristic in an organism is represented by two factors (it means that each cell has two chromosomes, carrying the gene for the same character).
- When two contrasting factors are present in an organism then one of them can mask the presence of the other. Therefore, one is called the **dominant factor**, while the other is called the **recessive factor**.
- When two contrasting factors are present in an individual, they do not blend and produce an intermediate type. However, they remain separate and get expressed in the F_2 progeny. The plant with Tt genotype is tall and not of intermediate height.
- When more than two factors are involved, these are independently inherited.

Mendel's Laws of Inheritance

Based on his experiments, Mendel proposed three laws or principles of inheritance-

- Law of Dominance
- Law of Segregation
- Law of Independent Assortment

Law of dominance and law of segregation are based on monohybrid cross while law of independent assortment is based on dihybrid cross.

Law of Dominance

- According to this law, characters are controlled by discrete units called factors, which occur in pairs with one member of the pair dominating over the other in a dissimilar pair.
- This law explains expression of only one of the parental character in F_1 generation and expression of both in F_2 generation.

Law of Segregation

- This law states that the two alleles of a pair segregate or separate during gamete formation in such a way that a gamete receives only one of the two factors.
- In homozygous parents, all gametes produced are similar; while in heterozygous parents, two kinds of gametes are produced in equal proportions.

Law of independent Assortment

- When two pairs of traits are combined in a hybrid, one pair of character segregates independent of the other pair of character.
- In a dihybrid cross between two plants having round yellow (RRYY) and wrinkled green seeds (rryy), four types of gametes (RY, Ry, rY, ry) are produced. Each of these segregate independent of each other, each having a frequency of 25% of the total gametes produced.

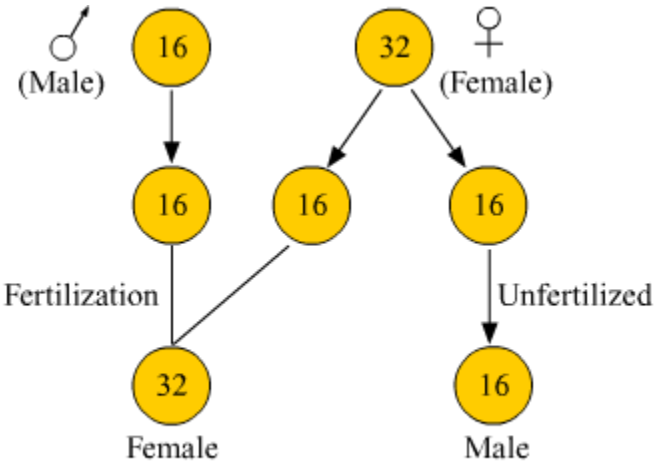
Sex Determination

- Henking discovered the genetic/chromosomal basis of sex determination by working on insects. He observed specific nuclear structures during spermatogenesis in insects. He named these structures as X bodies.
- He observed that after spermatogenesis, 50% of the sperm obtained these structures, while 50% did not.
- Later on, it was found that the X body observed by Henking was actually a chromosome and thus, this chromosome was named X chromosome.
- Chromosomes involved in sex determination are called sex chromosomes, while the other chromosomes are called autosomes.
- XO type of sex determination
- Other than autosomes, at least one X chromosome is present in all insects.
- Some sperms contain X chromosomes, while some do not.
- Eggs fertilised by sperms having X chromosomes become females. So, females have two X chromosomes.
- Eggs fertilised by sperms not having X chromosomes become males. So, males have only one X chromosome.
- Example of organisms with XO type of sex determination − Insects

- XY type of sex determination
- Males have X chromosome and its counterpart Y chromosome, which is distinctly smaller. Hence, males are XY.
- Females have a pair of X chromosomes. Hence, females are XX.
- Example of organisms with XY type of sex determination − Humans and *Drosophila*
- Male heterogamety − XO and XY types of sex determination are examples of male heterogamety.
- In XO type, some gametes have X chromosomes, while some gametes are without X chromosomes.
- In XY type, some gametes have X chromosomes, while some gametes have Y chromosomes.
- Female heterogamety − ZW type of sex determination is an example of female heterogamety.
- In ZW type, the female has one Z and one W chromosome, while the male has a pair of Z chromosomes.

Sex Determination in honeybees -

- Honey bees show a special mechanism of sex determination called the haplo-diploidy.
- In honeybees, the sex of the offspring is determined by the fertilization or non-fertilization of eggs, rather than the presence or absence of sex chromosomes.
- The unfertilized honey bee eggs normally develop into male progeny and are haploid in nature (have just one set of chromosomes).
- The fertilized honey bee eggs, differentiate into queens and worker bees and are diploid in nature (have two sets of chromosomes).



Sex determination in honeybees

What is Sex Linked Inheritance?

Genes carried by sex chromosome are said to be sex linked. The appearence of a trait because of the presence of an allele either on X chromosome or Y chromosome is called Sex-linked Inheritance.

Diseases observed in X-linked Inheritance

Any disease that is determined by the sex chromosomes, or that occurs due to defects in a gene on the sex chromosomes, is said to be sex linked. These diseases can descend to the offsprings from the parents through gametes. The diseases that occur due to any defective gene present on X chromosomes are known as X-linked diseases.

Most of these diseases are recessive in nature, that means, in the case of females, the defective allele should be present on both of the X chromosomes.

These disorders are more commonly observed in males as they have only a single X chromosome. A single recessive gene on that X chromosome will cause the disease. Most commonly observed diseases are:

- Haemophilia - It is a genetic disorder under which the sufferer (recessive X bearing male and homozygous recessive female) is at a risk of excessive blood loss leading to death as blood fails to clot.
- Colour blindness - It is also a genetic disorder in which the sufferer is unable to identify or distinguish between various colours.

The following example will explain the sex-linked inheritance of colour-blindness in humans more clearly.

Normal mother (XX) **Colour-blind father** (X°Y)

Colour-blind father (X°Y)

Sperms (two types)

Normal mother (XX) Eggs (All similar)	**X°**	**Y**
X	XX°	XY
X	XX°	XY

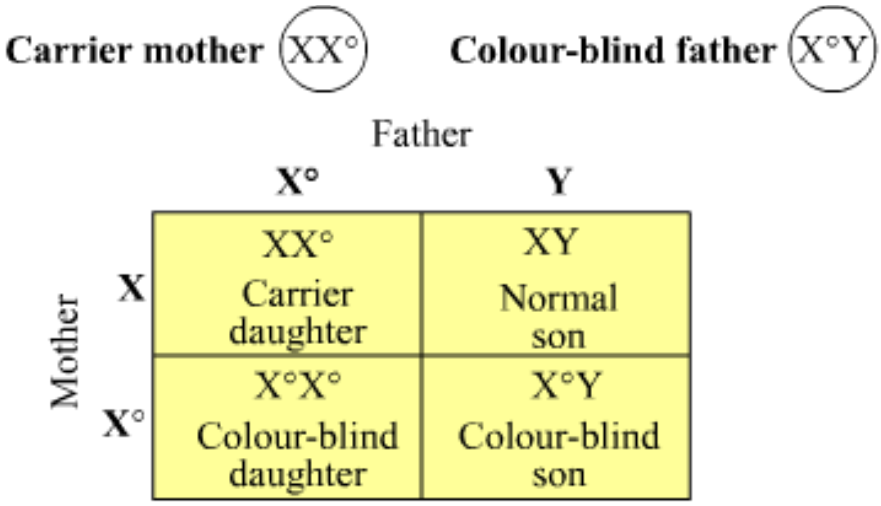
XX° : Daughters - heterozygous dominant, normal vision

XY : Normal sons

Carrier mother (XX°) **Normal father** (XY)

Normal father (XY)

Carrier mother (X°X) (not colour-blind)	**X**	**Y**
X°	X°X Carrier daughter	X°Y Colour-blind son
X	XX Normal daughter	XY Normal son



Criss-Cross Inheritance

The transfer of a gene from mother to son or father to daughter is known as criss-cross inheritance. For example, as in X-chromosome linkage.