Principles of Inheritance and Variation - Part 1

Objectives

After going through this lesson, the learners will be able to understand the following:

- Inheritance, Variations and Genetics.
- Important terms used in Genetics.
- About Mendel's Life.
- Nature of work done by Mendel.
- Mendel's Laws of Inheritance.

Content Outline

- Introduction
- Inheritance, Heredity and Variations
- Life History of Mendel
- Nature of Mendel's Work
- Why Did Mendel Choose a Pea Plant for His Experiments?
- Important Terms Used in Genetics
- Inheritance of One Gene
- Tools of Genetics (Punnett Square)
- Mendel's Laws of Inheritance
- Summary

Introduction

Have you ever wondered why an elephant always gives birth only to a baby elephant and not some other animal? Or why a mango seed forms only a mango plant and not any other plant? Given that they do, are the offspring identical to their parents? Or do they show differences in some of their characteristics? Have you ever wondered why siblings sometimes look so similar to each other? Or sometimes even so different?

These and several related questions are dealt with, scientifically, in a branch of biology known as Genetics. Genetics is the study of **genes**. Genes are made up of molecules inside the nucleus of a cell that are strung together in such a way that the sequence carries information: that information determines how living organisms inherit phenotypic traits (features) determined by the genes they received from their parents and thereby going back through the generations.

For example, offspring produced by sexual reproduction usually look similar to each of their parents because they have inherited some of each of their parents' genes. Genetics identifies which features are inherited, and explains how these features pass from generation to generation.

Some phenotypic traits can be seen, such as eye colour while others can only be detected, such as blood type or intelligence. Traits determined by genes can be modified by the surrounding environment: for example, the general design of a tiger's stripes is inherited, but the specific stripe pattern is determined by the tiger's surroundings. Another example is a person's height: it is determined by both genetics and nutrition.

Inheritance, Heredity and Variations

Inheritance

The transmission of a gene from parent to child. The pattern of inheritance is the manner in which a gene is transmitted.

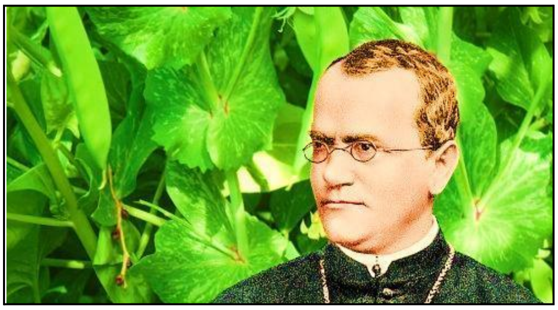
Heredity

Is the genetic information passing for traits from parents to their offspring, either through asexual reproduction or sexual reproduction. This is the process by which an offspring cell or organism acquires or becomes predisposed to the characteristics of its parent cell or organism.

Variations

Though offspring receive all the characters from their parents, they are not exactly alike. Differences are found even between the offspring of the same parents. It is difficult to find identical individuals. The progeny differs not only in itself but also with the parents. These differences are called **variations**. Thus, **variations** may be defined as **the visible differences between the parents and the offspring's or between the offspring's of the same parents**.

Gregor Mendel



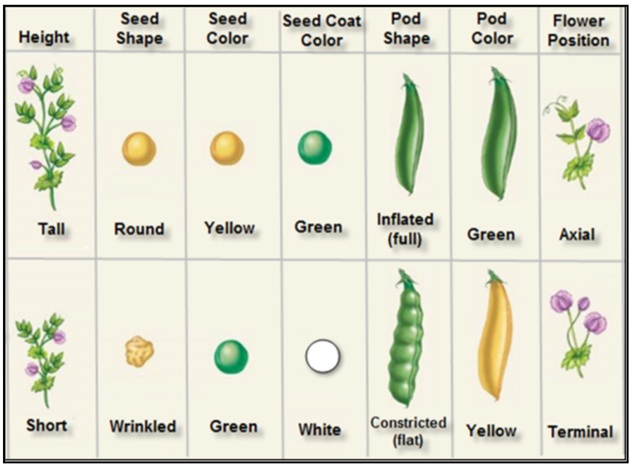
Mendel (1822-1884) was an Austrian monk at Brno monastery. He was a keen gardener and scientist, and studied at Vienna University, where he learnt statistics. Mendel's monastery had a 5 acre (2 hectare) garden, and his two former professors encouraged Mendel to pursue his interest in heredity by using the garden for experiments. He investigated inheritance in pea plants and published his results in 1866. They were ignored at the time, but were rediscovered in 1900, and Mendel is now recognized as the "Father of Genetics".

Nature of Mendel's work

After initial experiments with pea plants, Mendel settled on studying seven traits that seemed to inherit independently of other traits: seed shape, seed colour, flower colour, flower location, pod shape, unripe pod colour, and plant height. He first focused on seed shape, which was either wrinkled or round.

Between 1856 and 1863 Mendel cultivated and tested some 28,000 plants, majority of which were pea plants (***Pisumsativum)***.

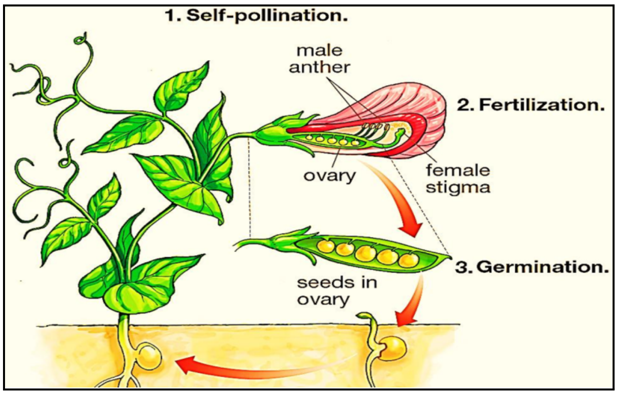
During Mendel's investigations into inheritance patterns it was for the first time that



statistical analysis and mathematical logic were applied to problems in biology. His experiments had a large sampling size, which gave greater credibility to the data that he collected. Also, the confirmation of his inferences from experiments on successive generations of his test plants, proved that his results pointed to general rules of inheritance rather than being unsubstantiated ideas. His experiments led him to make two generalizations, the **Law of Segregation** and the **Law of Independent Assortment**, which later came to be known as Mendel's Laws of Inheritance.

His experiments succeeded because:

- Mendel investigated simple well-defined characteristics (or traits), such as flower colour or seed shape, and he varied one trait at a time. Previous investigators had tried to study many complex traits, such as human height or intelligence.
- Mendel used an organism whose sexual reproduction he could easily control. Peas can also be self-pollinated, allowing self-crosses to be performed. This is not possible with animals.
- Mendel repeated his crosses hundreds of times and applied statistical tests to his results.
- Mendel studied two generations of peas at a time.



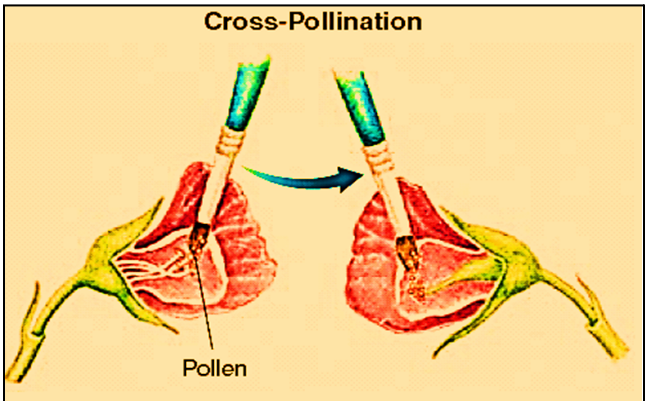
Why Did Mendel Choose a Pea Plant for His Experiments?

Mendel choose the pea plants for his experiments with the following reasons:

1. Normally the pea plant was self-pollinating (fertilizing), because petals enclose the reproductive organs till fertilization. The self-fertilization through many generations helps in easily obtaining the **pure line** with constant traits in pea plants.
2. The pea plant was easy to cultivate and from one generation to next took only a single growing season (annual).
3. Peas had many sharply defined inherited characters. Thus they possess many desirable features.

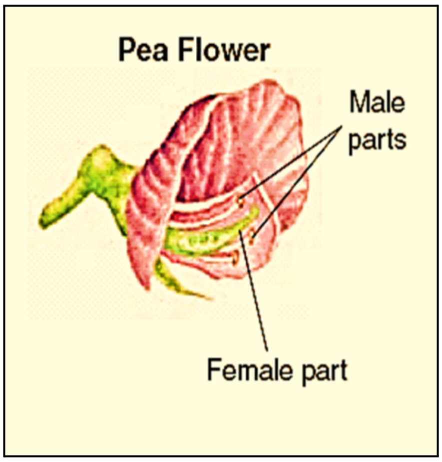
Characters	Contrasting Traits
Stem height	Tall/dwarf
Flower colour	Violet/white
Flower position	Axial/terminal
Pod shape	Inflated/constricted
Pod colour	Green/yellow
Seed shape	Round/wrinkled
Seed colour	yellow/Green

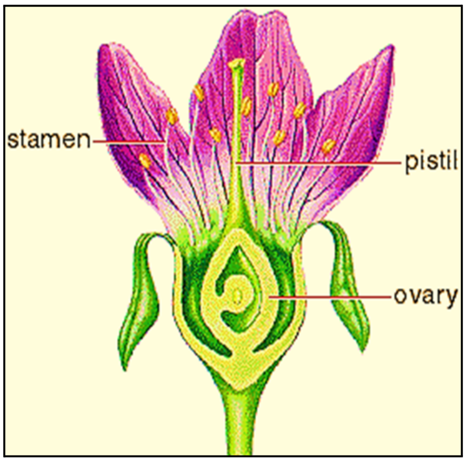
4. Cross pollination and fertilization can also be achieved easily.



Cross-Pollination To cross-pollinate pea plants, Mendel cut off the male parts of one flower and then dusted it with pollen from another flower.

5. The flowers are bisexual and hermaphrodite.





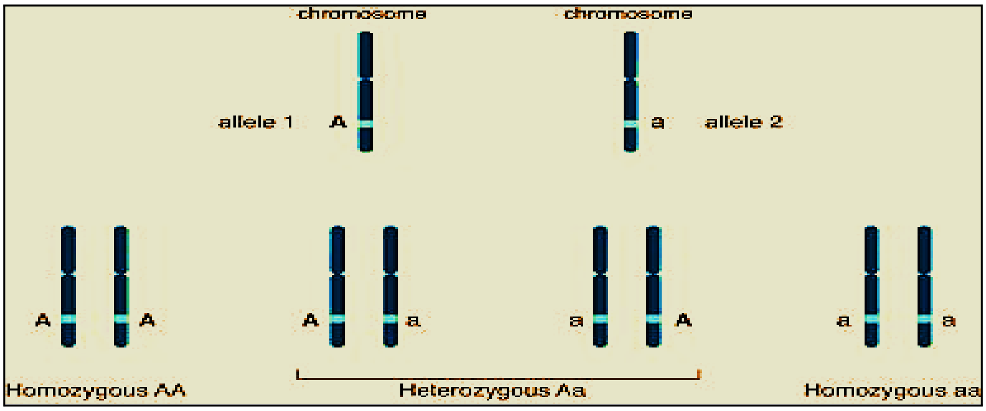
Important Terms Used in Genetics

Gene

A gene is a locus (or region) of DNA which is made up of nucleotides and is the molecular unit of heredity. A gene is a region of DNA that encodes function. A chromosome consists of a long strand of DNA containing many genes. A human chromosome can have up to 660 million base pairs of DNA with thousands of genes.

Allele

Genes exist in more than one form. These alternative forms of genes are called **alleles** and there are typically two alleles for a given trait. They are slightly different forms of the same gene. Allele determines distinct traits that can be passed on from parents to offspring.



Phenotype

A **phenotype** is the composite of an organism's observable characteristics or traits, such as its morphology, development, biochemical or physiological properties. A phenotype results from the expression of an organism's genes as well as the influence of environmental factors and the interactions between the two.

Genotype

The **genotype** is the part (DNA sequence) of the genetic makeup of a cell, and therefore of an organism or individual, which determines a specific characteristic (phenotype) of that cell/organism/individual. The genotype of an organism is the inherited instructions it carries within its genome.

Example

Alleles available: R and W

Possible genotypes:

RR, RW, WW

Dominant and Recessive Alleles

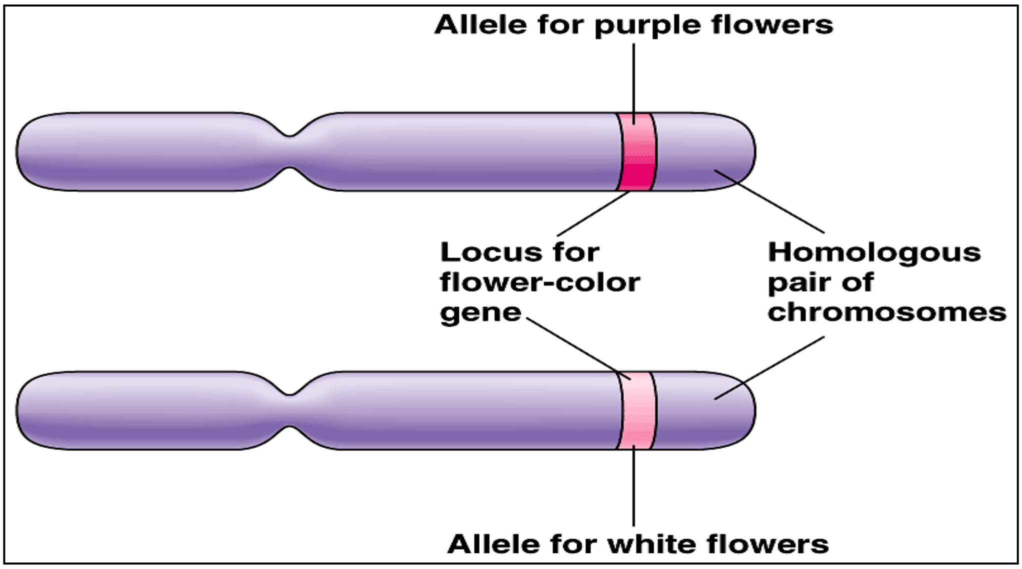
<u>Dominant alleles</u>:

"Dominate" over the other alleles

Will expresses itself in the presence of recessive, while a recessive allele is suppressed.

<u>Recessive alleles</u>:

Alleles that are suppressed in the presence of a dominant allele.



The purple-flower allele and white-flower allele are two DNA variations at the flower-colour locus

Homozygous

Homozygous refers to having identical alleles for a single trait. An allele is a form of a gene. Alleles can exist in alternative forms and organisms typically have two alleles for a given trait. Diploid cells have two alleles per trait, with one allele being donated from each parent. Sometimes the term "PURE" is used instead of homozygous.

Example: The gene for seed shape in pea plants exists in two forms, one form or allele for round seed shape (R) and the other for wrinkled seed shape (r). The round seed shape is dominant and the wrinkled seed shape is recessive. A homozygous plant would contain the following alleles for seed shape: (**RR**) or (**rr**). The (**RR**) genotype is homozygous dominant and the (**rr**) genotype is homozygous recessive for seed shape.

Heterozygous

Heterozygous refers to having two different alleles for a single trait.

A heterozygous genotype can also be referred to as **HYBRID**.

Example: The gene for seed shape in pea plants exists in two forms, one form or allele for round seed shape (R) and the other for wrinkled seed shape (r). A heterozygous plant would contain the following alleles for seed shape: **(Rr)**.

F_1 and F_2 generations

F_1 generation

The first generation is produced by a cross and consists of individuals heterozygous for characters in which the parents differ and are homozygous — also called first filial generation.

F_2 generation

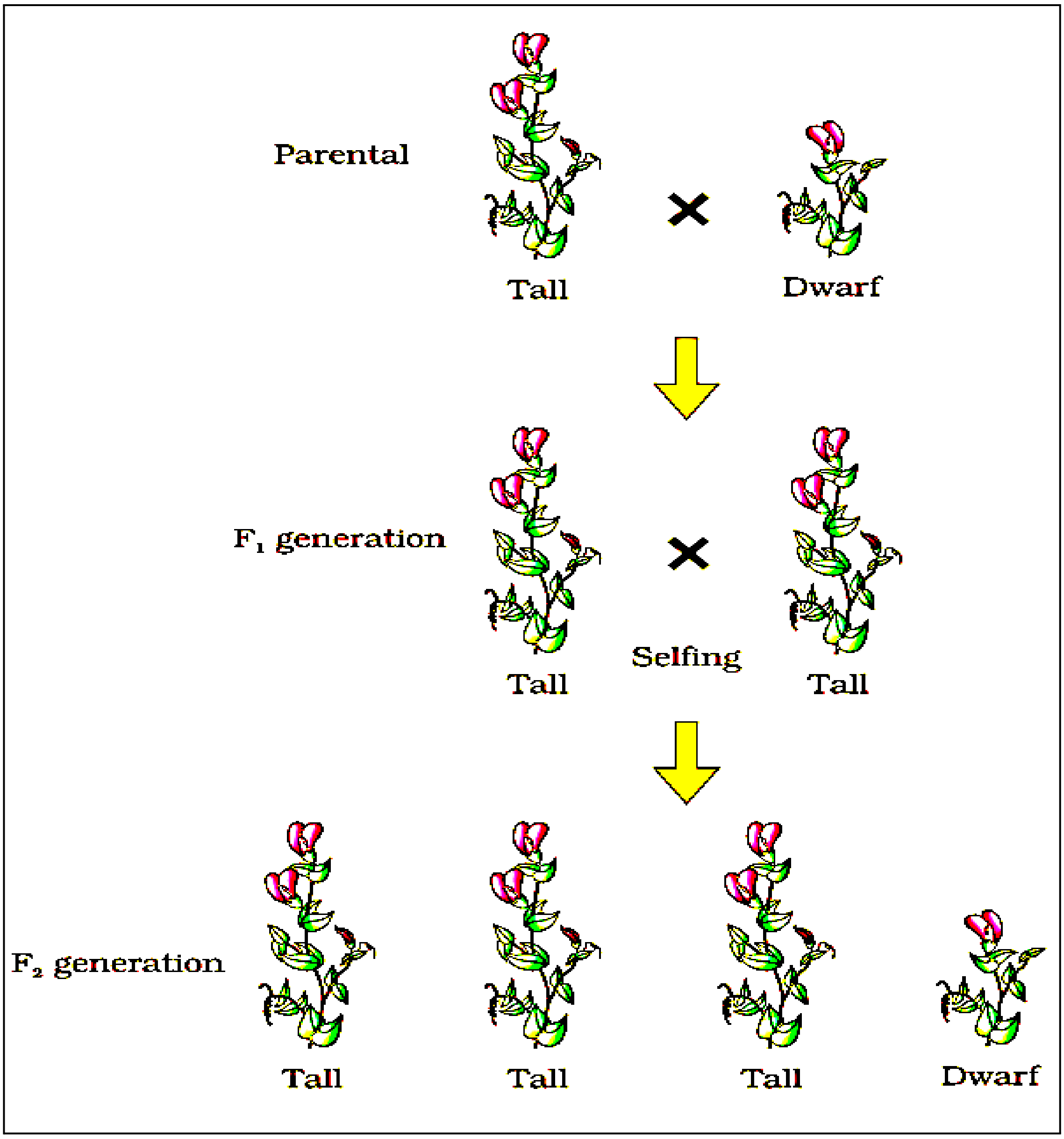
The second filial generation, which is composed of offspring(s) resulting from a cross of the members of F_1 generation.

Inheritance of One Gene

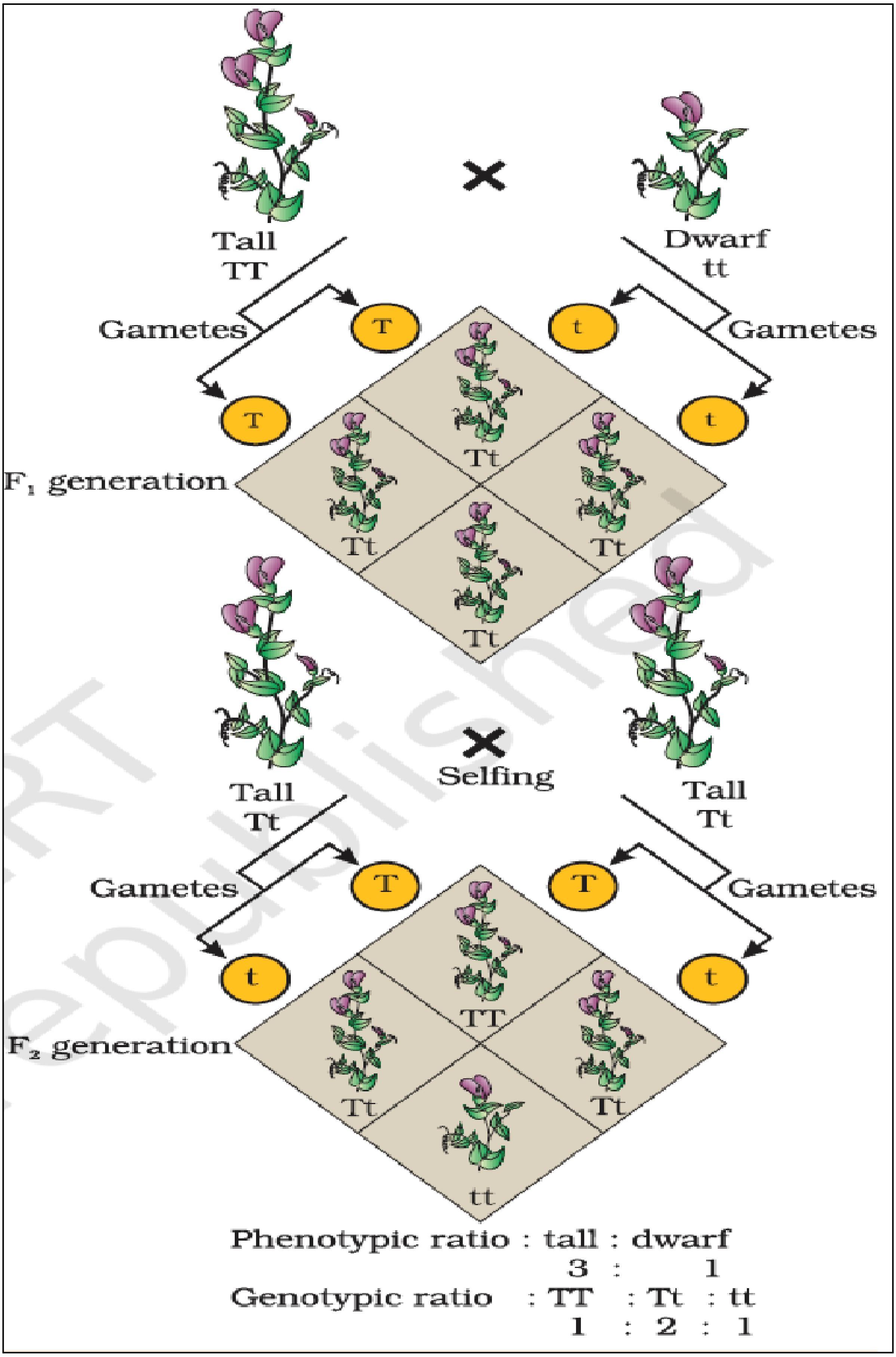
Let us take the example of one such hybridisation experiment carried out by Mendel where he crossed tall and dwarf pea plants to study the inheritance of one gene. He collected the seeds produced as a result of this cross and grew them to generate plants of the first hybrid generation. This generation is also called the Filial 1 Progeny or the F_1. Mendel observed that all the F_1 progeny plants were tall, like one of its parents; none were dwarf. He made similar observations for the other pairs of traits – he found that the F_1 always resemble either one of the parents, and that the trait of the other parent was not seen in them.

Mendel then self-pollinated the tall F_1 plants and to his surprise found that in the Filial 2 generation some of the offspring were '**dwarf**'; the character that was not seen in the F_1 generation was now expressed. The proportion of plants that were dwarf were $1/4^{th}$ of the F_2 plants while $3/4^{th}$ of the F_2 plants were tall. The tall and dwarf traits were identical to their parental type and did not show any blending, that is all the offspring were either tall or dwarf, none were of in between height. Similar results were obtained with the other traits that he studied:

Only one of the parental traits was expressed in the F_1 generation while at the F_2 stage both the traits were expressed in the proportion 3:1. The contrasting traits did not show any blending at either F_1 or F_2 stage.



The **Punnett Square** shows the parental tall TT (male) and dwarf tt (female) plants, the gametes produced by them and the F_1 Tt progeny. The F_1 plants of genotype Tt are self-pollinated. The F_1 plants of the genotype Tt when self-pollinated, produce gametes of the genotypeT and t in equal proportion. When fertilisation takes place, the pollen grains of genotype T have a 50 percent
chance to pollinate eggs of the genotype T, as well as of genotype t. Also pollen grains of genotype t have a 50 percent chance of pollinating eggs of genotype T, as well as of genotype t. As a result of random fertilisation, the resultant zygotes can be of the genotypes **TT, Tt or tt**.


Tall
TT
Dwarf
tt
Gametes
T
T
t
t
Gametes
F1 generation
Tt
Tt
Tt
Tt
Tall
Tt
Selfing
Tall
Tt
Gametes
T
t
T
t
Gametes
F2 generation
TT
Tt
Tt
tt
Phenotypic ratio : tall : dwarf
3 : 1
Genotypic ratio : TT : Tt : tt
1 : 2 : 1

Tools of Genetics

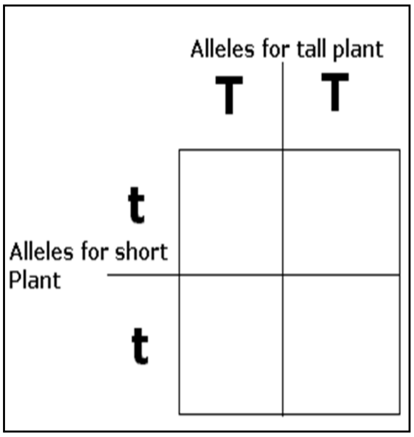
Punnett Square

The Punnett square is a diagram that is used to predict an outcome of a particular cross or breeding experiment. It is named after Reginald C. Punnett, who devised the approach. The diagram is used by biologists to determine the probability of an offspring having a particular genotype. The Punnett square is a tabular summary of possible combinations of maternal alleles with paternal alleles. These tables can be used to examine the genotypic outcome probabilities of the offspring of a single trait (allele), or when crossing multiple traits from the parents.

It is a tool for calculating genetic probabilities.

Punnett Process

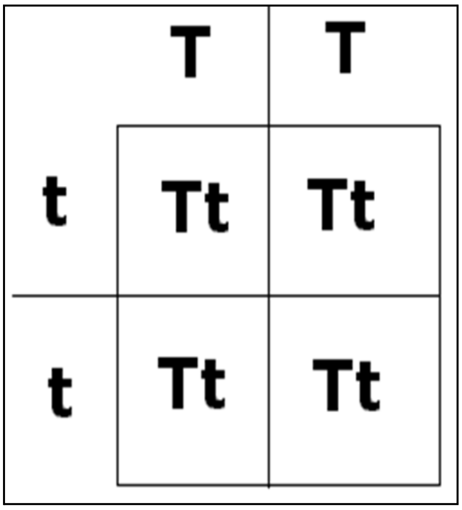
The diagram is used by biologists to determine the probability of an offspring having a particular genotype.



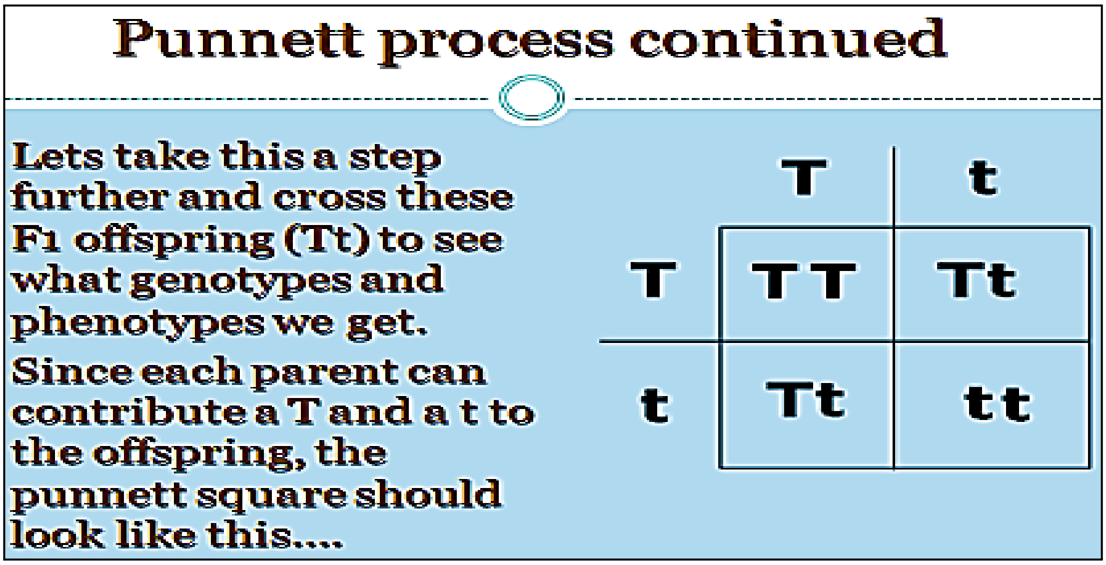
Punnett process

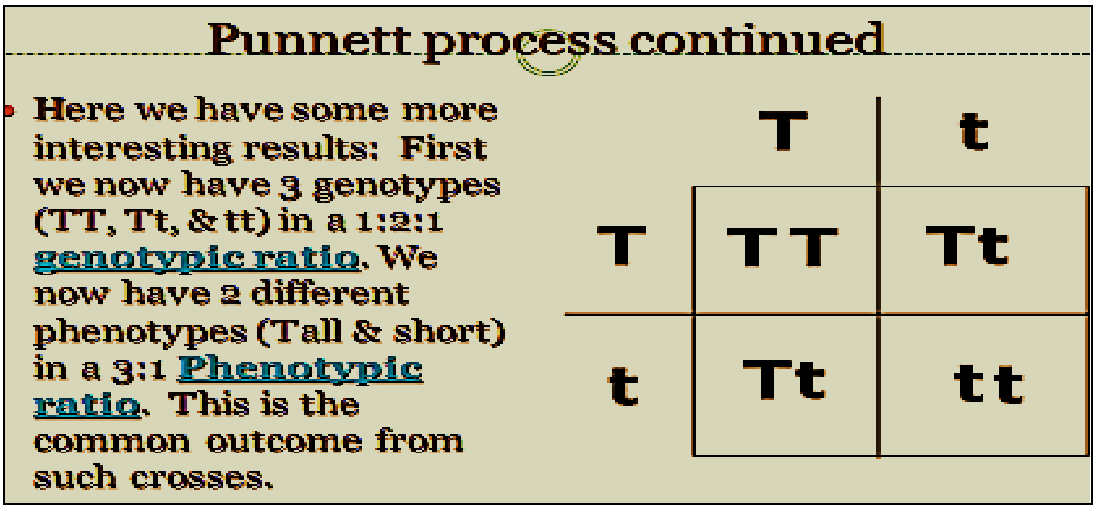
Determine alleles of each parent, these are given as TT, and tt respectively.

Take each possible allele of each parent, separate them, and place each allele either along the top, or along the side of the Punnett square.



Punnett process continued Lastly, write the letter for each allele across each column or down each row. The resultant mix is the genotype for the offspring. In this case, each offspring has a Tt (heterozygous tall) genotype, and simply a "Tall" phenotype.





Mendel's Laws of Inheritance

Mendel's Laws of Inheritance are as follows:

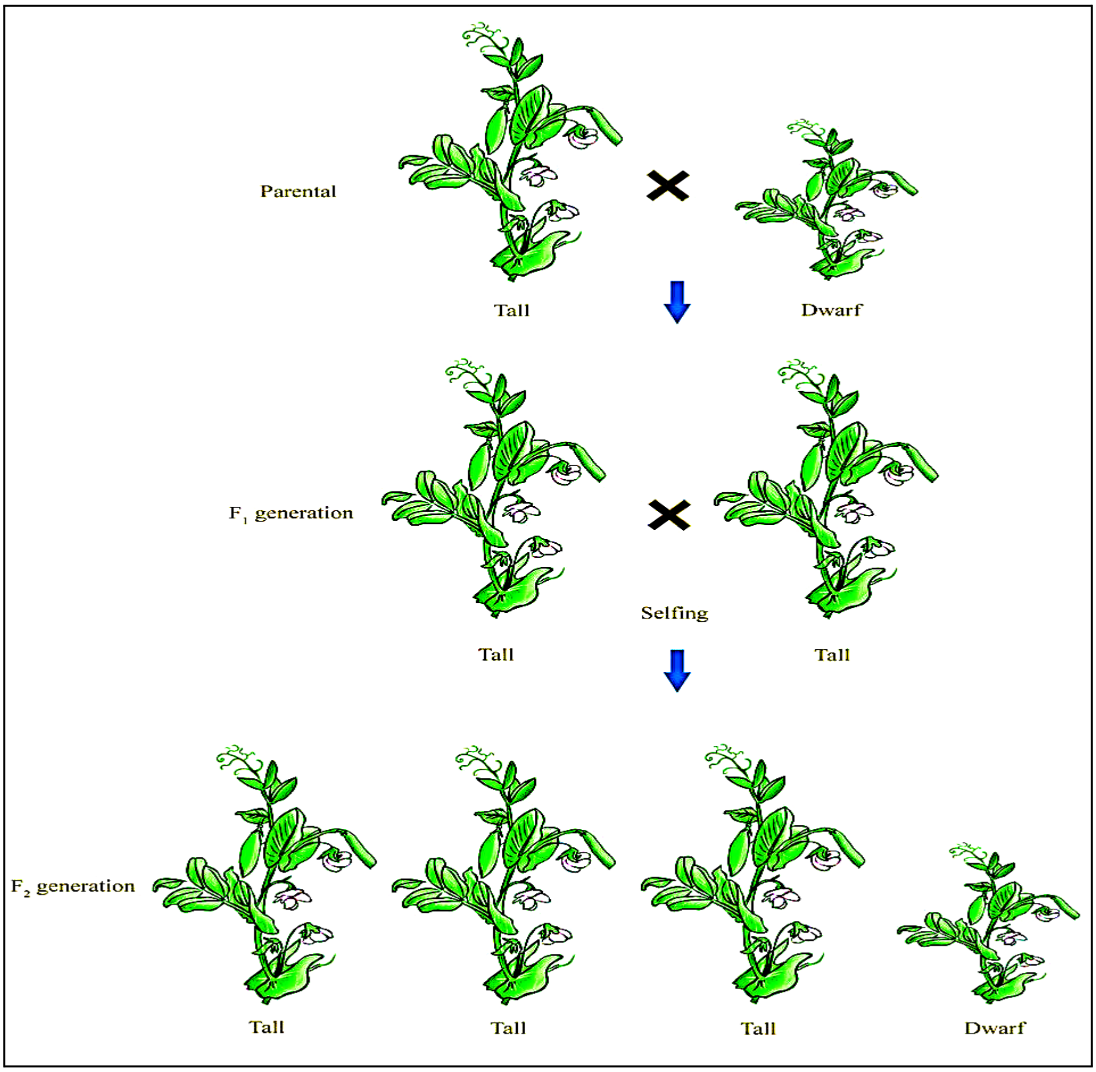
- The Law of Dominance
- The Law of Segregation
- The Law of Independent Assortment

The Law of Dominance (MonoHybrid Cross)

In a cross of parents that are pure for contrasting traits, only one form of the trait will appear in the next generation. Offspring that are hybrid for a trait will have only the dominant trait in the phenotype.

While Mendel was crossing (reproducing) his pea plants (over & over & over again), he noticed something interesting. When he crossed pure tall plants with pure short plants, all the new pea plants (referred to as the F1 generation) were tall. Similarly, crossing pure yellow seeded pea plants and pure green seeded pea plants produced an F1 generation of all yellow seeded pea plants. The same was true for other pea traits:

Parent Pea Plants	F1 Pea Plants
tall stem x short stem	all tall stems
yellow seeds x green seeds	all yellow seeds
green pea pods x yellow pea pods	all green pea pods
round seeds x wrinkled seeds	all round seeds
axial flowers x terminal flowers	all axial flowers



Summary

Genetics is a branch of biology which deals with principles of inheritance and its practices. Progeny resembling the parents in morphological and physiological features has attracted the attention of many biologists. Mendel was the first to study this phenomenon systematically. While studying the pattern of inheritance in pea plants of contrasting characters, **Mendel** proposed the principles of inheritance, which are today referred to as '**Mendel's Laws of Inheritance**'.

He proposed that the 'factors' (later named as **genes**) regulating the characters are found in pairs known as alleles. He observed that the expression of the characters in the offspring follow a definite pattern in different–first generations (F_1), second (F_2) and so on. Some characters are dominant over others. The dominant characters are expressed when factors are in heterozygous condition **(Law of Dominance)**. The recessive characters are only expressed in homozygous conditions.