

## 1. Details of Module and its structure

Module Detail	
Subject Name	Biology
Course Name	Biology 03 (Class XII, Semester - 1)
Module Name/Title	Principles of Inheritance and Variation – Part 2
Module Id	lebo_10502
Pre-requisites	Knowledge about Terms used in Genetics and idea of cross
Objectives	After going through this lesson, the learners will be able to understand the following: <ul style="list-style-type: none"><li>• Mendel's second law of inheritance.</li><li>• Reasons for Mendel's success.</li><li>• Concept of incomplete dominance.</li><li>• Concept of co-dominance.</li><li>• Test cross.</li><li>• Pleiotropy</li></ul>
Keywords	Gene, Allele, Dominant, Recessive, Homozygous, Heterozygous, Genotype, Phenotype, Punnett's Square, F1 and F2 generations or progenies.

## 2. Development Team

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## Introduction

The laws of inheritance were derived by Mendel, a nineteenth-century (1822-1884) Austrian monk conducting hybridization experiments in garden peas (*Pisum sativum*) (between 1856 and 1863). From these experiments, he induced two generalizations which later became known as *Mendel's Principles of Heredity*. He described these principles in a two-part paper, (*Experiments on Plant Hybridization*), that he read to the Natural History Society of Bruno on 8 February and 8 March 1865, and which was published in 1866.

Unfortunately Mendel's conclusions were largely ignored and not appreciated by contemporary biologists. Although they were not completely unknown to biologists of the time, they were not seen as generally applicable, even Mendel himself, was not sure of his findings, as he failed to get similar results on **hawkweed** (*Hieracium*) and thought they only applied to certain categories of species or traits. In fact His work was ahead of his time and he lacked aggressiveness in his personality. His mathematical approach in working out of biological problems was strange for the then scientists. He published his results in a journal which had a limited circulation.

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In 1900, however, his work was "**re-discovered**" by three European scientists, **Hugo de Vries, Carl Correns, and Erich von Tschermak**.

Regardless, the "re-discovery" made Mendelism an important but controversial theory. Its most vigorous promoter in Europe was William Bateson, who coined the terms "**genetics**" and "**allele**".

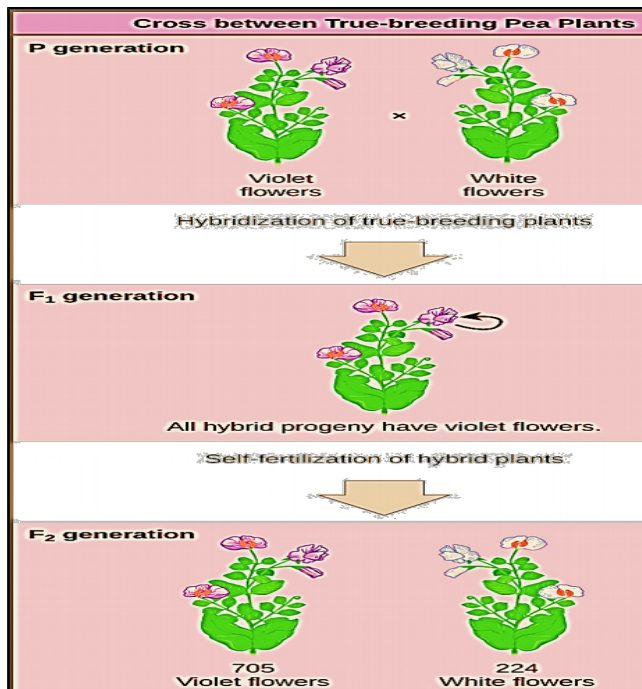
A large contribution to Mendel's success can be traced to his decision to start his crosses only with plants he demonstrated were true-breeding. His method of data analysis and his large sample size gave credibility to his data. He also had the foresight to follow several successive generations ( $F_2$ ,  $F_3$ ) of pea plants and record their variations. Finally, he performed "test crosses" (back-crossing descendants of the initial hybridization to the initial true-breeding lines) to reveal the presence and proportion of recessive characters.

### **Laws of Inheritance**

1. The Law of Dominance (already discussed in first Module)
2. The Law of Segregation (to be discussed in this Module)
3. The Law of Independent Assortment. (to be discussed in third Module)

### **Law of segregation:**

The Law of Segregation states that every individual organism contains two alleles for each trait, and that these alleles segregate (separate) during meiosis such that each gamete contains only one of the alleles. An offspring thus receives a pair of alleles for a trait by inheriting homologous chromosomes from the parent organisms: one allele for each trait from each parent.

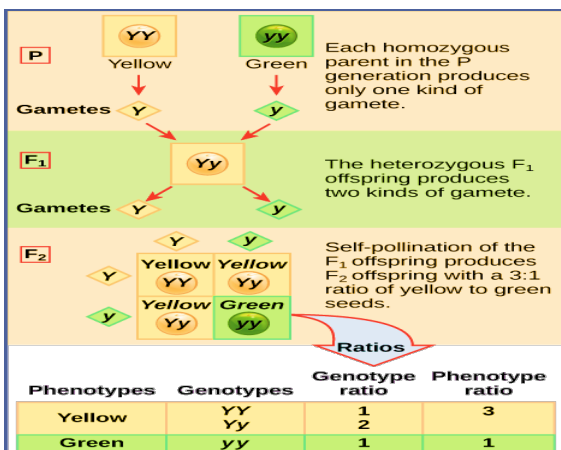


Since the gametes possess one allele of each character, they are always pure. The law of segregation is therefore also called the **law of purity of gametes**.

We now know that the segregation of genes occurs during meiosis in eukaryotes, which is a process that produces reproductive cells called gametes.

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It helps explain why progeny do not always resemble their parents. For example, two pea plants



with **yellow colour of seeds**, with the genotype **Yy** can be crossed to produce a pea plant with **green colour of seeds**. The two alleles, **Y** and **y**, do not mix or change each other even though they are present in the same individual. Each allele can contribute to the next generation.

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### Reasons of Mendel's success:

Some of the reasons for Mendel's success are:

- Mendel concentrated in one or few characters at a time.
- He made controlled crosses and kept careful numerical records of results.

- Mendel suggested 'factors' as the cause of characters
  - The experimental material, *Pisum sativum* was a wise choice
- Pisum sativum* has large bisexual flowers. As the flowers are large, emasculation is easy.

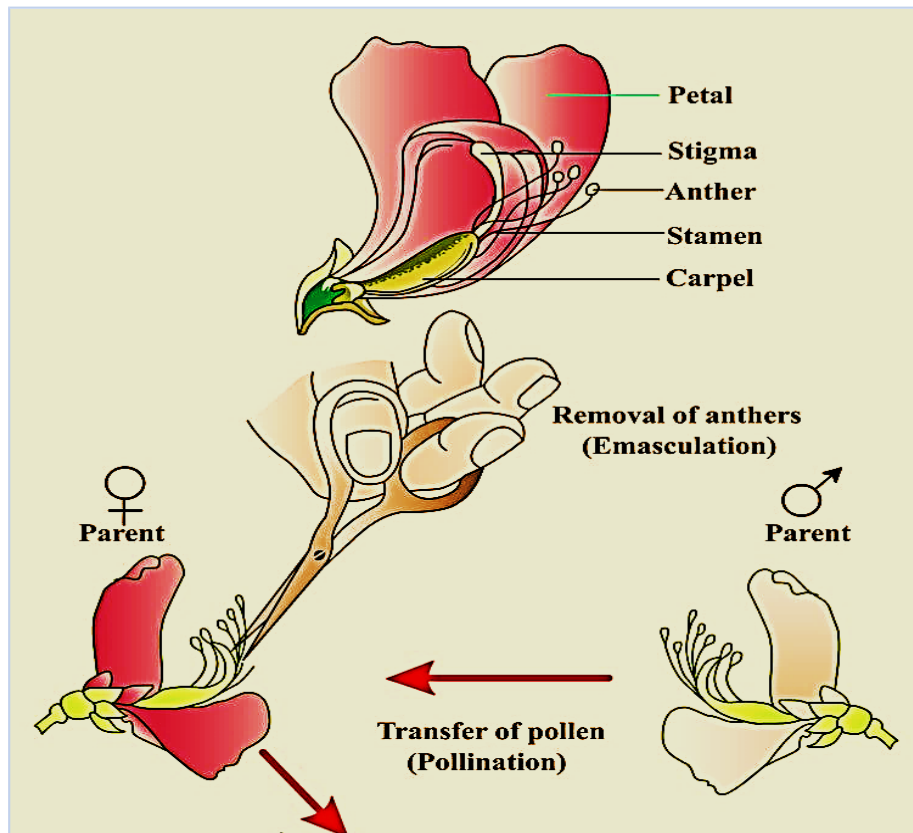


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Emasculation is the removal of anthers from bisexual flowers before their stigma become mature. Emasculation is a prerequisite for hybridization because pollination of a flower with its own pollen has to be totally prevented.



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- Garden pea is a naturally self-pollinated crop. Even then, hybridization with other varieties creates no problems like reduction in the number of seeds.

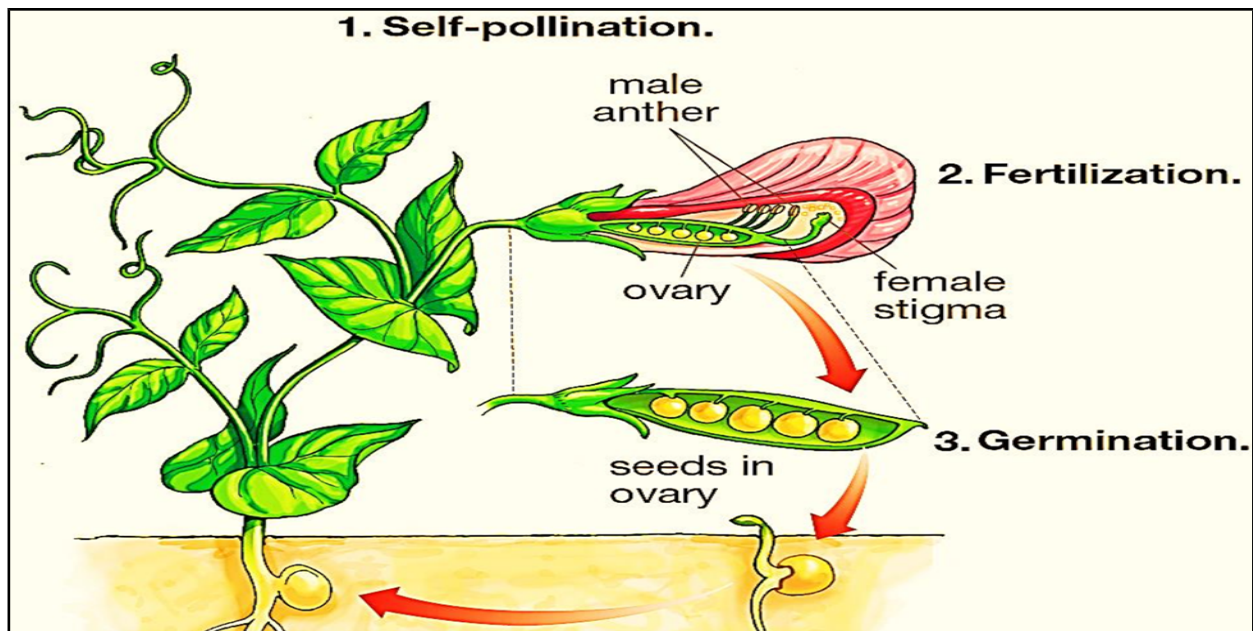


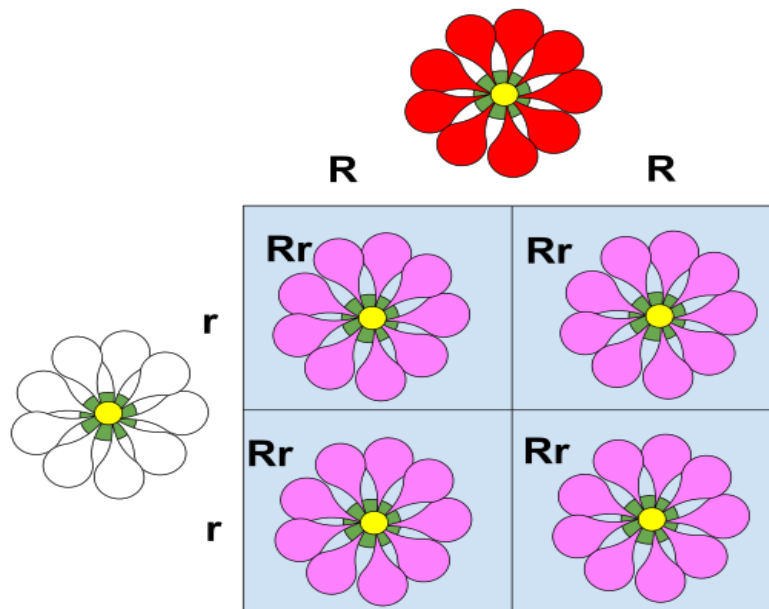
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- Many varieties are available in garden pea. So, any number of hybridization can be done taking two varieties at a time.
- As garden pea is short duration crop, three or four generation can be raised in a year. Thus, the course of transmission of factors through several generations can be traced within a short span of time.

## Incomplete dominance

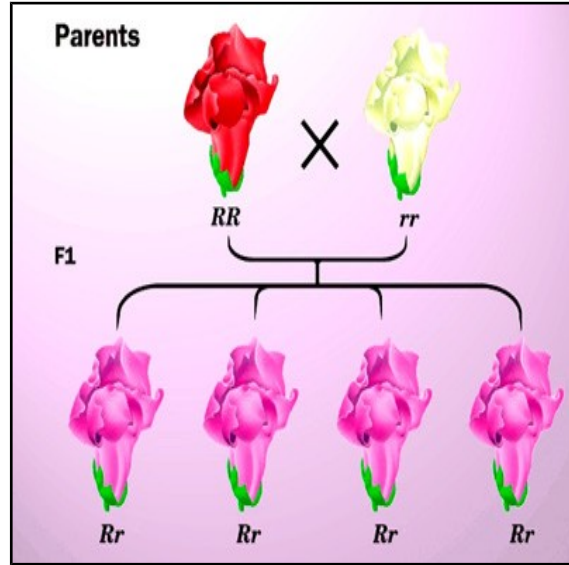
Incomplete dominance (also called partial dominance or semi-dominance) occurs when the phenotype of the heterozygous phenotype is distinct from and often intermediate to the phenotypes of the homozygous phenotypes.

For example, the snapdragon flower colour is either homozygous for red or white. When the red homozygous flower is paired with the white homozygous flower, the result yields a pink snapdragon flower. The pink snapdragon is the result of incomplete dominance.



A similar type of incomplete dominance is found in the four o'clock plant wherein pink colour is produced when true-bred parents of white and red flowers are crossed. In quantitative genetics, where phenotypes are measured and treated numerically, if a heterozygote's phenotype is exactly between (numerically) that of the two homozygotes, the phenotype is said to exhibit no dominance at all, i.e. dominance exists only when the heterozygote's phenotype measure lies closer to one homozygote than the other.





When plants of the F1 generation are self-pollinated, the phenotypic and genotypic ratio of the F2 generation will be 1:2:1 (Red:Pink:White).



Incomplete dominance in Snapdragon

Another example of incomplete dominance is sickle cell anemia, a disease in which the hemoglobin protein is produced incorrectly and the red blood cells have a sickle shape. A person that is homozygous recessive for the sickle cell trait will have red blood cells that all have the



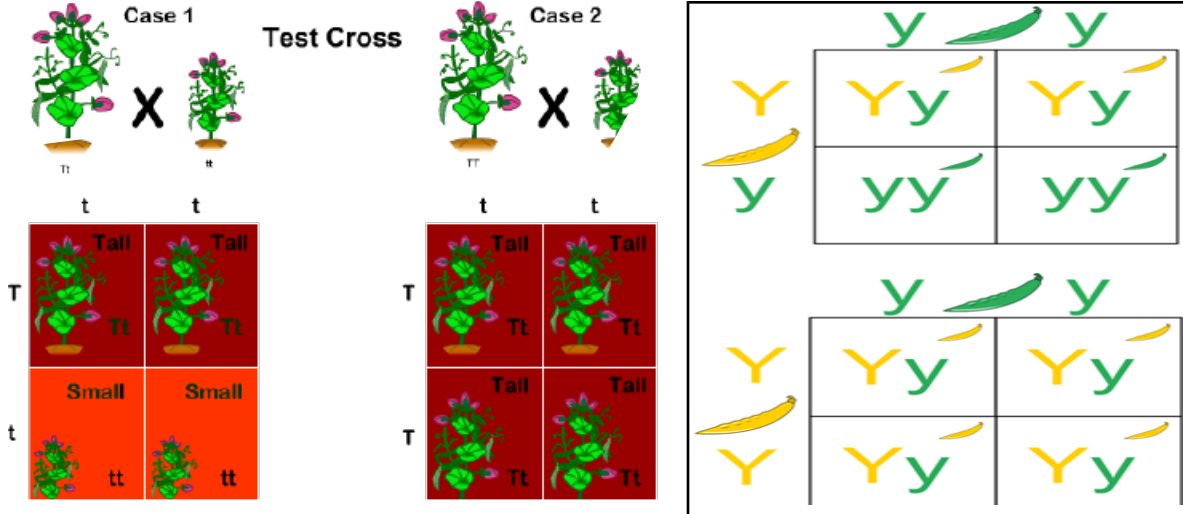
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incorrect hemoglobin. A person who is homozygous dominant will have normal red blood cells. And because this trait has an incomplete dominance pattern of expression, a person who is heterozygous for the sickle cell trait will have some misshapen cells and some normal cells. These heterozygous individuals have a fitness advantage; they are resistant to severe malaria. Both the dominant and recessive alleles are expressed, so the result is a phenotype that is a combination of the recessive and dominant traits.

**Test cross:**

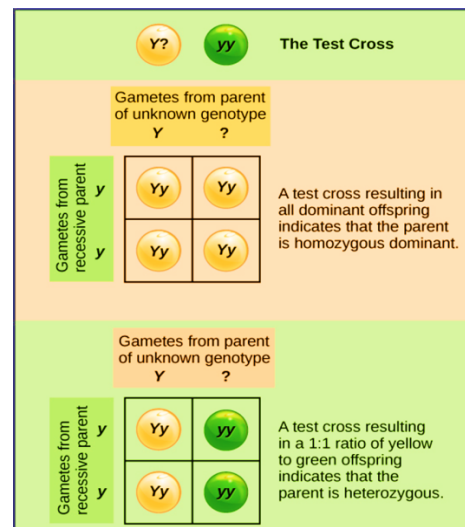
In genetics, a test cross, first introduced by **Gregor Mendel**, involves the breeding of an individual with a phenotypically recessive individual, in order to determine the zygosity of the former by analyzing proportions of offspring phenotypes. Zygosity can either be heterozygous or homozygous. Those that are heterozygous have one dominant and one recessive allele. Individuals that are homozygous dominant have two dominant alleles, and those that are homozygous recessive have two recessive alleles.

The genotype that an offspring has for each of its genes is determined by the alleles inherited from its parents. The combination of alleles is a result of the maternal and paternal chromosomes contributed from each gamete at fertilization of that offspring. During meiosis in gametes, homologous chromosomes experience genetic recombination and segregate randomly into haploid daughter cells, each with a unique combination of maternally and paternally coded genes. Dominant alleles will override the expression of recessive alleles.



Test crosses are used to test an individual's genotype by crossing it with an individual of a known genotype. Individuals that show the recessive phenotype are known to have a homozygous recessive genotype. Individuals that show the dominant phenotype, however, may either be homozygous dominant or heterozygous. The phenotypically dominant organism is the individual in question in a test cross. The purpose of a test cross is to determine if this individual is homozygous dominant or heterozygous.

Test crosses involve breeding the individual in question with another individual that expresses a recessive version of the same trait. Analyzing the proportions of dominant and recessive offspring determines if the individual in question is homozygous dominant or heterozygous. If all offspring from the test cross display the dominant phenotype, the individual in question is homozygous dominant; if half the offspring display dominant phenotypes and half display recessive phenotypes, then the individual is heterozygous. Since the homozygous recessive individual can only pass on recessive alleles, the alleles the individual in question passes on determine the phenotypes of the offspring.



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### Co-dominance:

Co-dominance occurs when the contributions of both alleles are visible in the phenotype.

A good example is different types of red blood cells that determine ABO blood grouping in human beings. ABO blood groups are controlled by the gene I. The plasma membrane of the red blood cells has sugar polymers that protrude from its surface and the kind of sugar is controlled by the gene. The gene (I) has three alleles  $I^A$ ,  $I^B$  and  $i$ . The alleles  $I^A$  and  $I^B$  produce a slightly different form of the sugar while allele  $i$  does not produce any sugar. Because humans are diploid organisms, each person possesses any two of the three I gene alleles.  $I^A$  and  $I^B$  are completely dominant over  $i$ , in other words when  $I^A$  and  $i$  are present only  $I^A$  expresses (because  $I$  does not produce any sugar), and when  $I^B$  and  $i$  are present  $I^B$  expresses.

But when  $I^A$  and  $I^B$  are present together they both express their own types of sugars: this is because of co-dominance. Hence red blood cells have both A and B types of sugars. Since there are three different alleles, there are six different combinations of these three alleles that are possible, and therefore, a total of six different genotypes of the human ABO blood types.

Allele from Parent 1	Allele from Parent 2	Genotype of offspring	Blood types of offspring
$I^A$	$I^A$	$I^A I^A$	A
$I^A$	$I^B$	$I^A I^B$	AB
$I^A$	$i$	$I^A i$	A
$I^B$	$I^A$	$I^A I^B$	AB
$I^B$	$I^B$	$I^B I^B$	B
$I^B$	$i$	$I^B i$	B
$i$	$i$	$i i$	O

## Multiple Alleles

Alleles are alternative forms of a gene, and they are responsible for differences in phenotypic expression of a given trait (e.g., brown eyes versus green eyes). A gene for which at least two alleles exist is said to be polymorphic. Instances in which a particular gene may exist in three or more allelic forms are known as multiple allele conditions. It is important to note that while multiple alleles occur and are maintained within a population, any individual possesses only two such alleles (at equivalent loci on homologous chromosomes).

A very good example of multiple-allele genes is the gene of ABO blood group system.

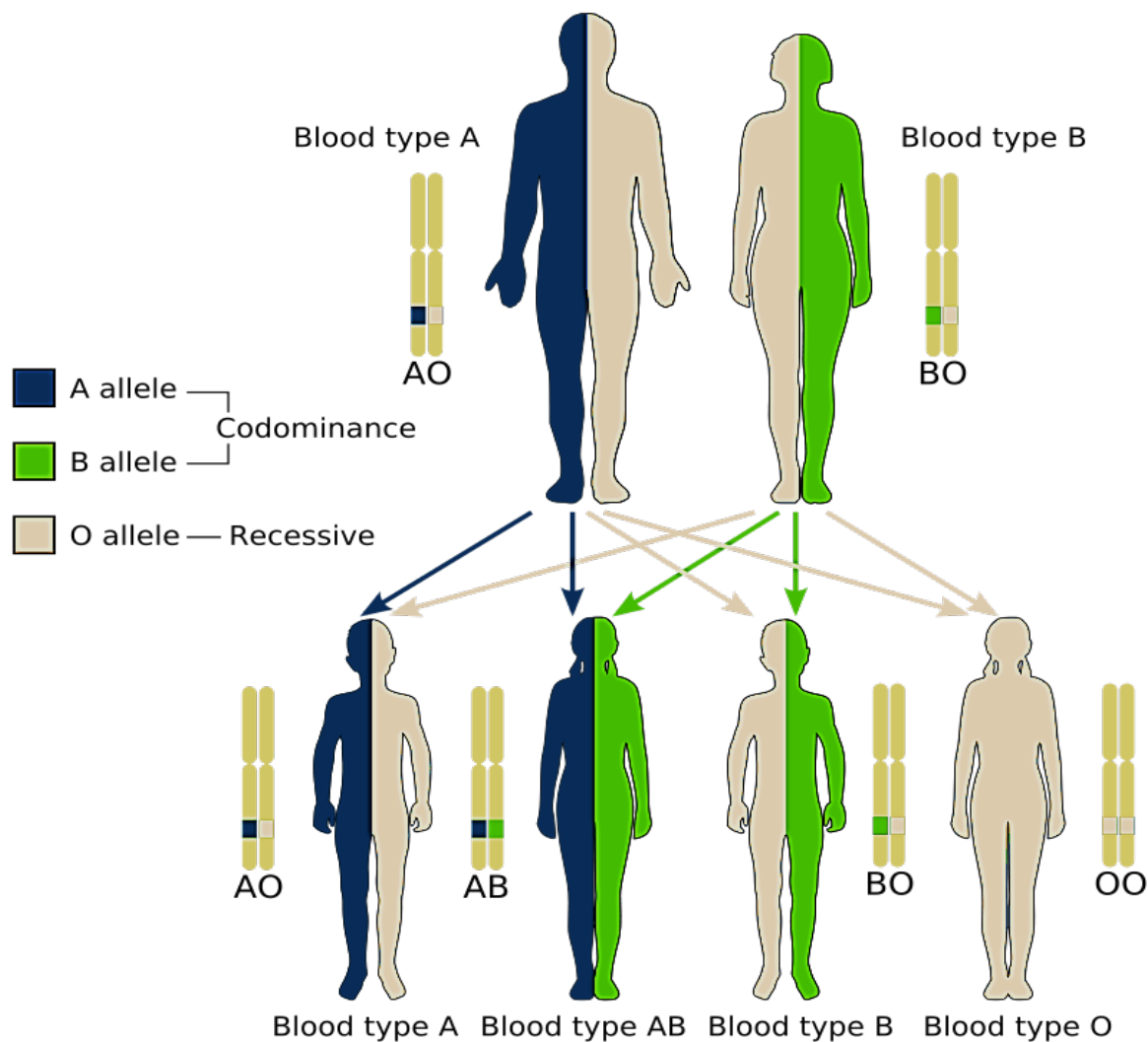


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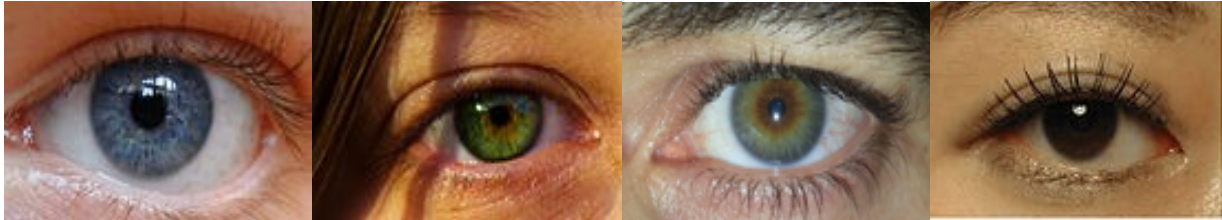
Co-dominance, where allelic products co-exist in the phenotype, is different from incomplete dominance, where the quantitative interaction of allele products produces an intermediate phenotype. For example, in co-dominance, a red homozygous flower and a white homozygous flower will produce offspring that have red and white spots. When plants of the F1 generation are self-pollinated, the phenotypic and genotypic ratio of the F2 generation will be 1:2:1 (Red:Spotted:White).



Co-dominance in a *Camellia* cultivar

Occasionally, a single gene product may produce more than one effect. For example, starch synthesis in pea seeds is controlled by one gene. It has two alleles (B and b). Starch is synthesised effectively by BB homozygotes and therefore, large starch grains are produced. In contrast, bb homozygotes have lesser efficiency in starch synthesis and produce smaller starch grains. After maturation of the seeds, BB seeds are round and the bb seeds are wrinkled. Heterozygotes produce round seeds, and so B seems to be the dominant allele. But, the starch grains produced are of intermediate size in Bb seeds. So if starch grain size is considered as the phenotype, then from this angle, the alleles show incomplete dominance.

Therefore, dominance is not an autonomous feature of a gene or the product that it has information for. It depends as much on the gene product and the production of a particular phenotype from this product as it does on the particular phenotype that we choose to examine, in case more than one phenotype is influenced by the same gene.



*Human eye colour is an inherited trait influenced by more than one gene. The interaction of multiple genes—and the variation in these genes ("alleles") between individuals—help to determine a person's eye colour phenotype. Eye colour is influenced by pigmentation of the iris and the frequency-dependence of the light scattering by the turbid medium within the stroma of the iris.*

## Summary

Mendel's laws of inheritance

**Law of dominance:** Some alleles are dominant while others are recessive; an organism with at least one dominant allele will display the effect of the dominant allele.

**Law of segregation:** During gamete formation, the alleles for each gene segregate from each other so that each gamete carries only one allele for each gene.

**Incomplete dominance:** It is a form of intermediate inheritance in which one allele for a specific trait is not completely expressed over its paired allele. This results in a third phenotype in which the expressed physical trait is a combination of the phenotypes of both alleles.

**Codominance:** Alleles that are masked or hidden by dominant alleles are known as recessive alleles. In some situations, both alleles are expressed equally. A genetic scenario where neither allele is dominant or recessive and both get expressed is known as codominance.

**Testcross:** A genetic cross between a homozygous recessive individual and a corresponding suspected heterozygote to determine the genotype of the latter.