Unit 8

Inheritance Biology

Unit Map

8.A Mendelian principles

The Mendelian principles are named after the Augustinian monk Gregor Johann Mendel (1822-1884, middle of the 19th century). He was considered as father of genetics due to his work on study of inheritance. He conducted cross breeding experiments on garden peas in his monastery garden in Brunn (Czech Republic) in 1865. Mendel recognized that hereditary is based on individual factors that are independent of each other. These factors are transferable from one generation to another in a particular pattern, each factor being responsible for an observable trait. The observable morphology of trait is known as phenotype and underlying genetic information is known as genotype. Mendel chose the garden pea, *Pisum sativum* as an experimental organism. Mendel chose seven traits as markers-flower colour, flower position, seed colour, seed shape, pod shape, pod colour and height.

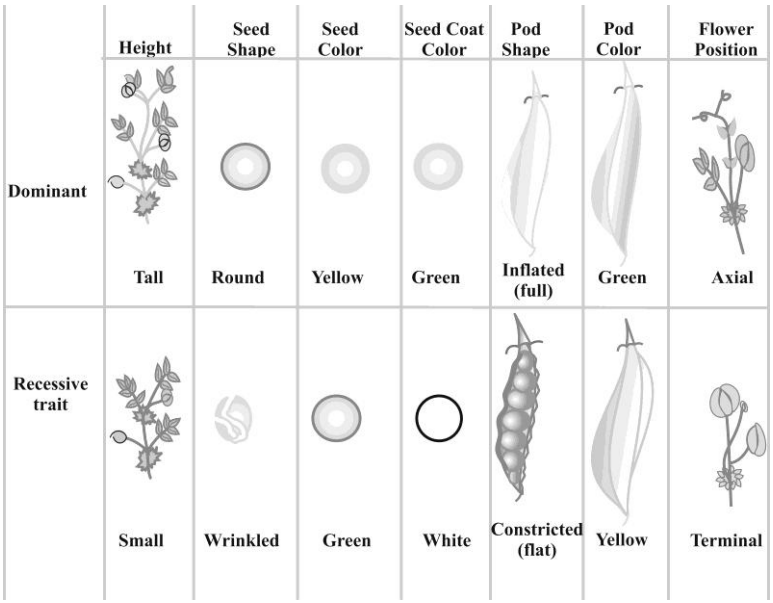


Figure 8.A.1-1
Figure showing basic seven traits of *Pisum sativum*, an experimental plant taken by Mendel to explain principles of genetics. Seven traits taken were height, seed shape, seed colour, seed coat colour, pod shape, pod colour and flower position. Each having two contrasting traits with one as dominant and one as recessive. Monohybrid and dihybrid crosses proved three principles of genetics. First is the law of dominance, according to which offspring will have traits of one parent because of dominance. Second is the law of segregation, two traits separate or segregate during reproduction and then combine in new combination in offspring. Third is law of independent assortment, according to which factors of characters assort independently without influencing others.

Mendel has done many monohybrid crosses using different variants of pea plant. Each of this crosses yielded approximately 3:1 ratio in the second generation (F2 generation). Three basic principles of inheritance were established or supported by Mendel monohybrid crosses.

8.A.1 Law of dominance

Firstly the progeny which he got in F1 generation had the appearance of one parent only this is because of dominance of one trait over the other one. This led him to propose law of dominance. According to this law when two dissimilar unit factors are present in an individual then only one expresses itself while other was completely masked. The one which expressed itself is said to be dominant and other which do not expressed itself is known as recessive.

8.A.2 Law of segregation

Mendel recognized that in order to get 3:1 ratio in the second generation each parent must possess two copies of the trait. This ratio makes him understand that the pea traits he was working with, has distinct units of heredity material. He proposed that these two traits separate or segregate during reproduction and then combine in new combinations in the offspring. This is known as law of segregation. Genotypic ratio 1:2:1

8.A.3 Law of independent assortment

In the di-hybrid cross, Mendel followed the same pattern as he did in the single-trait cross. The ratio 9:3:3:1 he obtained from di-hybrid cross was explained by him in the form of law of independent assortment. According to this law, the factors of different traits are not influenced by each other presence and each of the two factors of character can assort with either of two factors of another character. Genotypic ratio 1:2:2:1:4:1:2:2:1. The two characters considered were round and yellow (dominant) and wrinkled and green (recessive).

8.A.4 Deviation from Mendelian inheritance

- **Maternal inheritance**

  Topic discussed under unit 8.E.2

- **Epigenetic inheritance**

  Epigenetics is the study of heritable changes in gene activity that are not caused by changes in the DNA sequence.

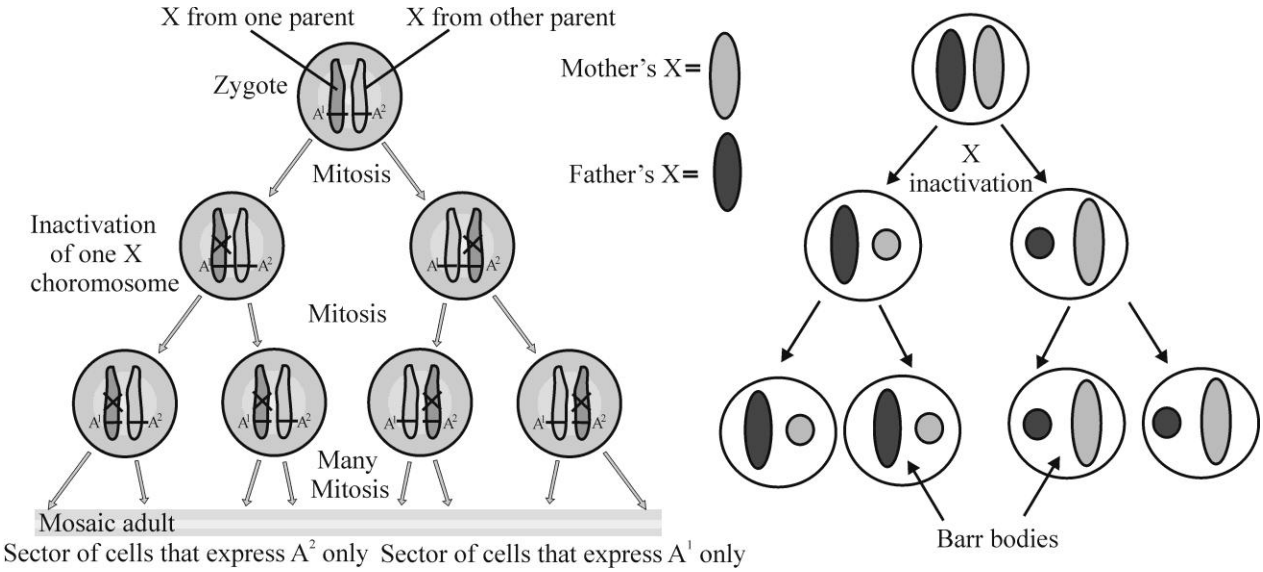
  Epigenetic inheritance is a pattern in which a nuclear gene or chromosome gets modified itself that changes the gene expression. This phenomenon is not permanent for many generations. These are the result of DNA and chromosomal modifications which takes place during oogenesis, spermatogenesis or early stages of embryogenesis. Once they are initiated during these early stages, these changes may change the expression of particular genes that may remain fixed during an individual's lifetime. Epigenetic changes may affect the phenotype of an individual permanently.

Two examples to be studied under this topic

i. **Genomic imprinting**

Topic discussed in unit 8.C.5

ii. **Dosage compensation** In 1961, Mary Lyon proposed that mammalian females have two X chromosomes and males have only one which led to the evolution of special mechanisms of dosage compensation. This phenomenon of inactivation of X chromosome in females equalizes the expression of genes in both sexes. This phenomenon of X- chromosome inactivation (XCI) is an extraordinary example of long range, monoallelic gene silencing and facultative heterochromatin formation and is also known as Lyon Hypothesis. In mammals, dosage compensation for X-linked gene products between XX and XY individuals is achieved by silencing one of the two X chromosomes in female cells. In 1949, Murray Barr and Ewart Bertram identified a highly condensed structure in the interphase nuclei of somatic cells in female cats that was not found in male cats. This structure was now recognized as the **Barr body.**



8.B Concept of gene

From Mendel work he proposed that each genetic character is controlled by a pair of unit "factors". The term factor used by Mendel implies to "gene" which was first used by Johansen in 1909. After him, many people worked on genes and proved that genes are part of DNA molecule.

T.M Morgan proposed the gene theory which state that

- Chromosomes are bearers of hereditary units and each chromosome carries hundreds or thousands of genes.
- The genes are arranged on the chromosome in the linear order and on the special regions or locus.

Classical concept of Gene

This concept was introduced by Sutton in 1902 and was further elaborated by Morgan in 1903.

The salient features of this concept are as follows

Genes are discrete particles inherited in Mendelian fashion that occupies a definite locus in the chromosome and are responsible for specific phenotypic expression. The number of genes is more than number of chromosomes hence several genes are located on each chromosome.

- The genes are arranged in a single linear order like beads on a string and each gene occupies specific position called ***locus.***
- Genes can be transmitted from parent to off springs.
- Genes are capable of combined together or can be replicated once during a cell division.
- Sudden changes in the position and composition of genes are called as ***mutation.***
- Genes are capable of self-duplication producing their own exact copies.

Modern Concept of gene

Benzer in 1957 coined different terms for different nature of gene and genetic material in relation to the chromosome on the basis of genetic phenomena to which they involve.

Following are the salient features of this concept of gene

- **Genes as cistrons** is the part of DNA that specifies a single polypeptide chain. It transmits characters from one generation to other as unit of transmission.
- **Genes as recon Recon** is a unit of recombination. It is the smallest segment of DNA capable of being separated and exchange with other chromosome.
- **Genes as Muton** is a unit of mutation. It is the smallest unit of genetic material which when changed or mutated produce a phenotypic trait.

8.B.1 Allele

An allele is one of two versions of a gene that is inherited from parents. If the two alleles which are inherited are same then that is referred to as homozygous and if alleles are different than it is referred as heterozygous.

8.B.2 Multiple Alleles

Generally one gene has only two alleles but in some cases one gene has more than two alleles. An excellent example of multiple allele is in heritance of human blood group type. Blood groups exist as four possible phenotypes: A, B, AB & O and these blood groups are controlled by one I gene. There are three alleles for the gene that determines blood type - I^A, I^B and I^O.

8.B.3 Pseudo-allele

Pseudo-allele is an allele that is functionally but not structurally allelic. It means that their wild type recombinants can be recovered by intragenic recombination from heterozygous containing two different alleles. Genes controlling Rh blood group are called Pseudo-alleles.

- **Chromosomal theory of inheritance**

  This theory was proposed by Sutton and Boveri independently in 1902.

  The salient features of this theory are as follows

  - The genes and both the chromosomes occur in pairs in the somatic or diploid cells.
  - A gamete contains only one chromosome of a type and only one of the two alleles of a character.
  - The paired condition of both chromosomes as well as Mendelian factors is restored during fertilization.

8.B.4 Complementation tests

Topic discussed under unit 8.I.3

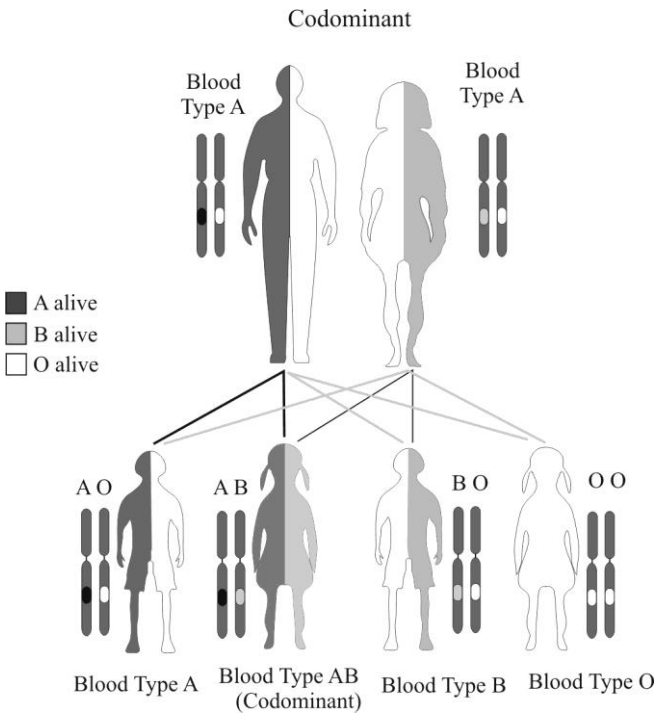
8.C Extension of Mendelian principles

8.C.1 Co-dominance

Co-dominance is defined as a type of Non-Mendalian inheritance pattern that finds the traits expressed by the alleles to be equal in the phenotype. There is neither a complete dominance nor incomplete dominance of one trait over the other for that given characteristic. Co-dominance would show both alleles equally instead of a blending of the traits as is seen in incomplete dominance.

In the case of co-dominance, the heterozygous individual expresses both alleles equally. There is no mixing or blending involved and each is distinct and equally shown in the phenotype of the individual. Neither trait masks the other like in simple or complete dominance.

Many times, co-dominance is linked with a characteristic that has multiple alleles. That means there is more than just two alleles that code for the trait. Some traits have three possible alleles that may combine and some traits have even more than that. Often times, one of those alleles will be recessive and the other two will be co-dominant. This gives the trait the ability to follow the Mendelian Laws of heredity with simple or complete dominance or, alternatively, to have a situation where co-dominance comes into play.



One example of co-dominance in humans is the AB blood type. Red blood cells have antigens on them that are designed to fight off other foreign blood types, which is why only certain types of blood can be used for blood transfusions based on the recipient's own blood type. A type blood cells have one kind of antigen, while the B type blood cells have a different type. Normally, these antigens would signal that they are a foreign blood type to the body and would be attacked by the immune system. People with AB blood types have both antigens naturally in their systems, so their immune system will not attack those blood cells.

This makes people with the AB blood type "universal recipients" due to the co-dominance displayed by their AB blood type. The A type does not mask the B type and vice - versa. Therefore, both the A antigen and B antigen are equally expressed in a display of co-dominance.

8.C.2 Incomplete dominance

Incomplete dominance is a form of intermediate inheritance in which one allele for a specific trait is not completely dominant over the other allele. This results in a third phenotype in which the expressed physical trait is a combination of the dominant and recessive phenotypes.

Incomplete dominance is similar to but different from co-dominance. In co-dominance, an additional phenotype is produced; however both alleles are expressed completely. Co-dominance is exemplified in AB blood type inheritance.