MENDELISM

Genetics is the study of genes, genetic variation, and heredity in living organisms.

The Austrian monk “Gregor John Mendel” is considered as the Father of "Modern Genetics". Mendel made experiments on garden pea plants, species of Lathyrus (**Pisum** **sativum**).

Johan Gregor Mendel conducted a number of experiments with pea plant in the kitchen garden of the parish. He observed several contrasting characters in pea plants such as a tall variety and a dwarf variety, yellow seeds and green seeds, round seeds and wrinkled seeds etc. These characters were handed down from generation to generation because the pea plants are self-pollinated. The seven contrasting characters that were taken into account by Mendel are as follow:

1. Seed shape - Round or wrinkled
2. Cotyledon color - Yellow or green
3. Seed coat color - Colored or white
4. Pod shape - Inflated or constricted
5. Pod color - Green or yellow
6. Flower position - Axial or terminal
7. Height - Tall or dwarf

Based on the observations of his experiments on garden pea, Mendel drew some important conclusions. These conclusions are known as Mendel's Laws of Inheritance. These are as follows:

1. Law of Dominance
2. Law of Segregation
3. Law of Independent Assortment

1. **Low of dominance**

'In monohybrid cross where the parents differ in one pair of contrasting characters the f1 generation shows only one trait which is termed as dominant, the other character which is hidden or not expressed is called recessive'

In **monohybrid crosses**, he observed that F1 offspring or monohybrids show characters or traits of only one parent. It simply indicates that out of two contrasting characters only one appears in the F1 generation and the other disappears.

This led him to formulate his first law of heredity the Law of Dominance, which states that: **"One character or factor prevents the expression of other".**

The characters which appear in the F1 generation are called dominant and those which do not appear are termed as recessive. This appearance of the dominant character in the F1 generation is termed as the law of dominance.

Example: Mendel crossed pure tall plants with the pure dwarf plants. The seeds thus obtained were sown which gave rise to tall plants:

Cross Tall x Dwarf - Parent generation (P1)

TT x tt

Tt Ist filial generation (F1)

(Tall)

Thus in F1 generation, only the tall (Tt - Dominant, hybrid) character appears which prevented the expression of the dwarf (tt - Recessive) character.

2. **Law of segregation**

Law of segregation is based on the results and observations of the **F2** generations of the monohybrid crosses. After the observing the results of F1 generation, Mendel experimented further and self-fertilized the flowers of **F1** plants or generation. The seeds thus obtained from these flowers were sown and developed into plants **(F2).** Mendel noted that all these plants were similar to the original plants i.e. **P1** generation and F1 plants. They were found to be in a ratio of 3 : 1 (3 plants showing dominant character and 1 showing recessive character). This led Mendel to formulate his second law which is called as **"Law of Segregation: or "Law of Purity of Gametes".** It states that:

"The hybrids or heterozygote of F1 generation contain two contrasting characters of dominant and recessive nature. These characters do not mix with each other but segregate or separate at the time of gamete formation in such a manner that each gamete receives only one character either dominant or recessive".

This law is also called as the law of purity of gametes because the gametes contain only one character and are pure for it.

Example: This law may be explained with an example of a garden pea. The hybrid of **F1** produced by the crossing of a homozygous (pure) tall plant and a homozygous (pure) dwarf plant was tall. The flowers of this tall plant on self-fertilization produced seeds which in **F2** generation developed into tall and dwarf plants in the ratio of **3:1**. Actually, Mendel obtained **787** tall and **277** dwarf plants. Their ratio is approximately **3:1.**

Cross - I

Tall x Dwarf ------------------- Parent generation (P1)

TT tt

Tt (Tall) -------------------- Ist filial generation (F1)

Cross II TT X tt

Gametes	**T**	**t**
T	**TT** (Tall)	**Tt** (Tall)
T	**Tt** (Tall)	**tt** (Dwarf)

3Tall : 1 Dwarf

1 TT : 2 Tt : 1 tt

Homozygous : heterozygous : homozygous

3. Law of Independent Assortment:-

After **monohybrid experiments Mendel** tried dihybrid crosses. For this **Mendel** crossed plants that differed in two characters. He crossed pea plant having yellow round seeds with the plant having green wrinkled seeds. In **F1** generation he obtained dihybrid which had yellow and round seeds (dominant hybrid).

But when he self-fertilized the plants developed by these seeds of **F1**, he did not find **3 : 1** ratio as was found in the monohybrid experiments. But in **F2** generation he found four types of seeds in the ratio of **9 : 3 : 3 : 1**. Out of four types of seeds, two types of seeds were like the original parents (**P1** generation) but two types quite new. They neither resembled the parents nor the hybrid of **F1**. By the observations of the dihybrid experiments, **Mendel** formulated his third law of **Independent Assortment.**

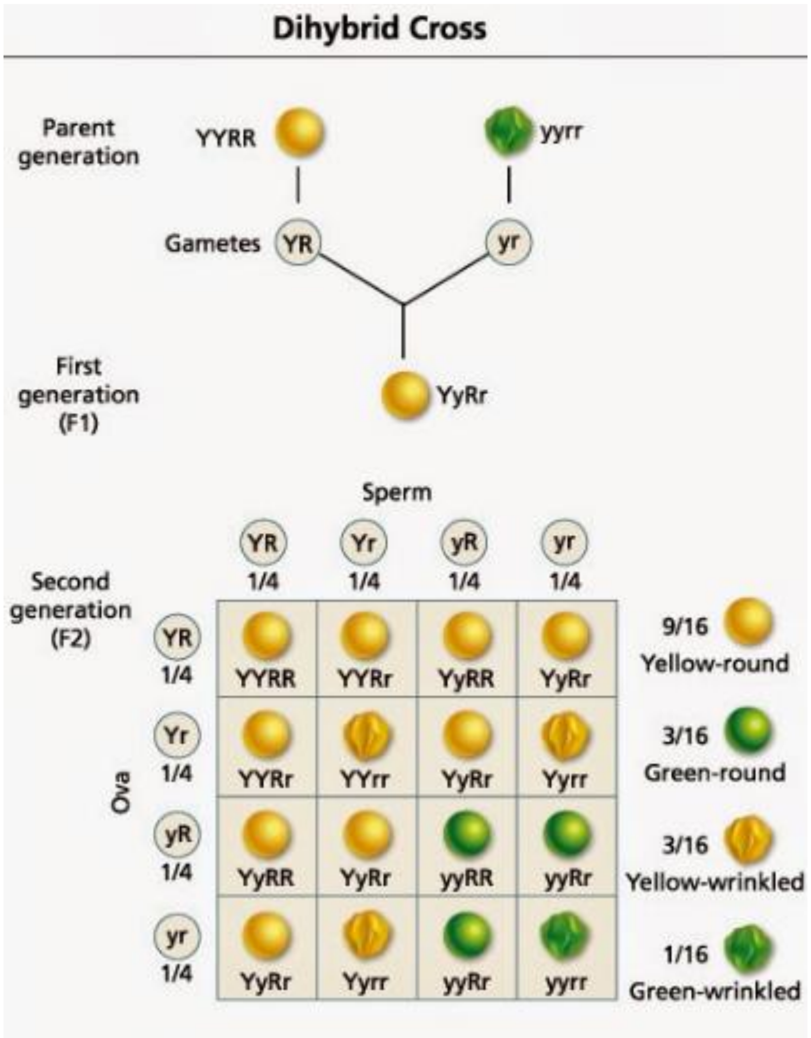
Mendel explained that the two characters (seed color and seed shape) are not tied together but they remain independent of each other. The round shape of the seed is not always associated with the yellow color; however, it may remain associated with the green color also. Consequent on these findings the law states that:

"The factors (now genes) for different pairs of contrasting characters segregate or assort independently of each other at the time of gametogenesis in F1 hybrid without affecting or diluting each other".

Example: When a pea plant having yellow and rounded seeds were crossed with the other having green and wrinkled seeds, in **F1** generation all the hybrid plants produced yellow and rounded seeds. When these seeds were sown, the plants developed, which were self-fertilized. After self-fertilization the plants produced **4 types** of seeds which appeared in the ratio of **9: 3:3: 1.**

1. Yellow round - 9
2. Yellow wrinkled -3

3. Green round - 3
4. Green wrinkled - 1



Phenotypic ratio 9 : 3 : 3 : 1

Yellow Round   Yellow Wrinkled   Green Round   Green Wrinkled.

Genotypic ratio 1 : 2 : 1 : 2 : 4 : 2 : 1 : 2 : 1

YYRR   YYRr   YYrr   Yy RR   YyRr   Yyrr   yyRR   yyRr   yyrr

BACKCROSS AND TESTCROSS

Backcross is the cross between the F1 hybrid and its parent. For example, in a monohybrid cross when all tall plant is crossed to a dwarf plant the F1 hybrid is tall. The F1 hybrid is crossed to the all tall parent or to the dwarf it is called a back cross.

Backcross Tt x TT tall plant (dominant parent)

Gamets T t T

Gametes	T	t
T	TT (tall)	Tt (tall)

All plants are tall

F1 generation **Tt** all tall plants

<u>Test cross</u> Tt x Tt dwarf (recessive parents)

Gametes T t t t

Gametes	T	t
t	Tt (tall)	Tt (tall)

Phenotypic ratio = Tall : Dwarf

1 : 1

Test cross

When the F1 generation is backcrossed with the recessive parent it is called a test cross. The term test cross is used because when the F1 hybrid is crossed with its recessive parent we can find out if the organism is homozygous or heterozygous. The result of attest cross in a monohybrid cross is always 1:1

Plant bearing yellow and round seeds   plant bearing green and wrinkled

YYRR   X   yyrr

↘ ↙

YyRr Yellow round

F1 generation   YyRr   **x**   YyRr

YR   Yr   yR   yr   **x**   YR

gametes	**YR**	**Yr**	**yR**	**yr**
YR	YYRR Yellow round	YYRr Yellow round	YyRR Yellow round	YyRr Yellow round

All plants bear only yellow and round seeds.

<u>Test cross</u>   **YyRr**   **x**   **YYRR**

Gametes   YR Yr   yR   yr   yr

Gametes	**YR**	**Yr**	**yR**	**yr**
Yr	YyRr Yellow round	Yyrr Yellow and wrinkled seeds	yyRr green and round seeds	yyrr green and wrinkled seeds

The phenotypic ratio is

Yellow and round : Yellow and wrinkled : Green and round : Green and wrinkled

1   :   1   :   1   :   1

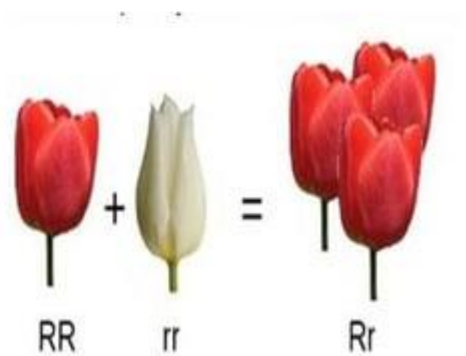
In a dihybrid cross the F1 hybrid is back crossed to its recessive parent it is called a test cross as we find out if the F1 organism is homozygous or heterozygous . The result of a test cross is always 1:1:1:1

Complete dominance

Complete dominance in genetics is when one allele completely dominates or takes control of the other. An **allele** is a version of a gene

Complete dominance is a form of dominance in the heterozygous condition where in the allele that is regarded as dominant completely masks the effect of the allele that is recessive. For instance, for an individual carrying two alleles that are both dominant (e.g. *RR*), the trait that they represent will be expressed. But if the individual carries two alleles in a manner that one is dominant and the other one is recessive, (e.g. *Rr*), the dominant allele will be expressed while the recessive allele will be suppressed. Hence, the heterozygote (*Rr*) will have the same phenotype as that of the dominant homozygote (*RR*). This condition is called **complete dominance**.

For instance, for an individual carrying two alleles that are both dominant (e.g. *RR red*) and individual carrying two alleles that are both recessive (e.g. *rr white*) only the dominant allele is getting expressed (Rr red)



Incomplete Dominance

The law of dominance status that the character that is seen in the F1 hybrid is the dominant one and the one that is hidden is recessive. In some cases a gene may be completely dominant or completely recessive. This concept is called incomplete dominance. In such cases the F1 hybrid exhibit an intermediate expression of both genes.

Incomplete dominance can be defined as the inability of the dominant gene to completely mask the expression of the recessive gene. In case of incomplete dominance the F1 generation hybrid will show a phenotypic character that is intermediate between the two contrasting characters. Thus the expressions of both genes are seen resulting in an intermediate phenotypic trait.

In case of **Mirabilis jalapa,** plants bear red fowers that are thought to be dominant over white flowers. The gene for red flowers is represented by '**RR'** and the genes for white flowers is represented by '**rr'.**

In a simple monohybrid cross a plant bearing red flowers is crossed with a plant bearing **white flowers**. The F1 generation will consist of plants bearing **pink flowers.** This is against the Law of Dominance where only the dominant trait is supposed to be expressed and the other trait, which is recessive, is hidden. The presence of pink flowers indicates that the gene for red flowers **'R' was not completely dominant over the gene for white flowers 'r' Or the recessive gene r is not completely recessive and cannot be completely masked by the gene 'R'** This is incompletely dominance.

When the F1 plants undergo self pollination, the F2 generation will comprise of three types of plants,

Plant bearing Red flower **RR** **x** **rr** plant bearing white flowers

Gametes R r

F1 generation Rr (pink flower)

gemetes	**R**	**r**
R	**RR** Red flower	**Rr** Pink flower
R	**Rr** Pink flower	**rr** white flower

F2 generation comprises of plants bearing **Red flower, pink flower. And white flower in the ratio 1:2:1**

PENETRANCE AND EXPRESSIVITY

A genotype produces a phenotype. Normally in a population if a particular dominant allele iss present in all individuals, the trait will be expressed in all of them. In some of them who possess the dominant allele may not express it or the trait is not seen.

Example – the gene for Huntington'disease

Let's assume that all members of a population have the dominant allele for Huntington's disease. If only **95%** of the population actually express it or are affected and 5% do not express any symptoms of the disease, the penetrance of the allele is **95%.**

Penetrance can be defined as**- the probability of gene or trait being expressed**

(In spite of the presence of a dominant allele, a phenotype may not be present or expressed)

Example – polydactyly in humans (extra fingers and /or toes). A dominant allele produces polydactyly in humans but not all humans with allele display the extra digits.

Complete and incomplete penetrance

If all individuals of a population who possess the gene or genes the trait, it called complete penetrance

'**Incomplete' or reduced penetrance means the genetic trait is expressed in only part of the population.**

The variation in penetrance may be due to a combination of genetic, environmental, and lifestyle factors, may of which are unknown. Some disease may express in only a certain age group.

<u>Expressivity</u>

Expressivity refers to variation in phenotypic expression when an allele is penetrant or expressed.

In polydactyly an extra digit may occur on one or more appendages. In some only one extra finger is present only in one hand. Some have an extra digit in both hands. In some people extra digits are seen in the legs.Here the allele show variation in expression.

Variation in expressivity refers to the range og sign and symptoms that can occur in different people with the same genetic condition.

Like reduced penetrance, variation in expressivity is probably caused by a combination of genetic, environmental, and lifestyle factors, most of which have not been identified.

If the digits can be full size or just a stub the allele has reduced penetrance as well as variable expressivity.