Genetics

- Genetics is the study of **heredity** and **variations.** Its expression influences the functions of individuals at all levels.
- Evidently, this branch of biology involves the study of molecules, cells, organisms and populations as well using many different experimental approaches.
- Genetic studies have many applications in the areas of agriculture and medicine.
- For example **Gene therapy** is used to replace the defective gene with normal gene in treating some diseases.
- Knowledge of genetic plays a significant role in understanding the processes of evolution.
- **Bateson** coined the term Genetics. Later Francis **Galton** coined the term **Eugenics** which is the direct application of genetic knowledge for human welfare. Later the term **Euphenics,** that deals with the symptomatic treatment of genetic disorders was coined.
- Although inheritance of biological traits had been recognised for thousands of years, the first fruitful results were obtained only by **Gregor Johann Mendel**, who is regarded as **'father of genetics'.**

MENDEL'S LAWS (TRANSMISSION GENETICS)

GREGOR JOHANN MENDEL

- **Johann Mendel** was born in 1822 and was admitted into Augustinian Monastery of St. Thomas in Brunn of Austria (Brno of Czech republic) in 1843 and took the name **Gregor.**
- From 1851 to 1853 he attended the University of Vienna, where he studied physics and botany.
- In 1865, he described his research in two lectures to the **Natural History Society** in Brunn.
- His work was published in a German Scientific Journal in1866 as a paper entitled "Experiments in Plant Hybridisation".

Mendel's experimental approach

- Mendel chose *Pisum sativum,* the garden pea plant for his experimental work.
- Mendel worked with seven "**characters**".
- Characters are the visible features that are represented by two **contrasting traits** (allelomorphic traits).
- Contrasting traits are the two alternative forms of a character.
- For example tall and dwarf are the contrasting traits of the character length of the stem in *Pisum sativum.* Mendel selected pure lines for his experimentation. A pure line is the one that produces the identical progeny on self pollination or self fertilization.
- On self pollination, a pure tall plant produces all tall plants and a dwarf plant produces only dwarf plants.

Character	Contrasting traits	F_1 results	F_2 results	F_2 ratio
Shape of the seed	round/wrinkled	all round	5474 round, 1850 wrinkled	2.96 : 1
Color of the cotyledon	yellow/green	all yellow	6022 yellow, 2001 green	3.01 : 1
Shape of the pod	full/constricted	all full	882 full, 299 constricted	2.95 : 1
Color of the pod	green/yellow	all green	428 green, 152 yellow	2.82 : 1
Position of the flower	axial/terminal	all axial	651 axial, 207 terminal	3.14: 1
Color of the flower **Color of the seed coat**	Gray/white	Gray	705 violet 224 white	3.15:1
Height of the stem	tall/dwarf	all tall	787 tall, 277 dwarf	2.84 : 1

The factor for colour of flower (violet/white) also influences the character colour of the seed coat. So, this factor is an example of pleiotropy (one factor influences)

- **Mendel selected *Pisum sativum* as his experimental material** for the following reasons:
  1. These plants are easy to cultivate and complete their life cycle in one season only.
  2. They have sharply distinct heritable differences, but Mendel has selected only seven characters.
  3. Method of pollination in these plants is self pollination. Therefore they are pure lines.
  4. The hybrids, produced are fertile.
  5. It is easy to conduct cross pollination in these plants, if required.
  6. Large number of offspring will be obtained. So experimental error can be minimised.
- Many scientists conducted hybridisation experiments before Mendel, but they were not successful in their experiments because they aimed at observing the inheritance of many characters in a single experiment.
- **Mendel's success** in the genetic experiments can be attributed to the following:
  1. Mendel restricted his experiments to one or a few pairs of contrasting traits in each experiment.
  2. Mendel, as he had mathematical background, kept accurate quantitative data, which is necessary in genetic experiments.
  3. Mendel also kept track of each character separately.
  4. *Pisum sativum* is with seven homologous pairs of chromosomes. The unit factors for the seven characters identified are located on different homologous pairs in this plant.
- Mendel formulated some postulates from his results that had become the principles of heredity.
- These postulates are later presented by Correns as genetic laws or Mendel laws - **the law of segregation** and **the law of independent assortment.**

Terminology in Genetics :

- New terminology is introduced to explain the Mendel's experiments.
  1. **Phenotype:** The physical appearance of a trait
  2. **Genotype:** The genetic makeup of an individual**.**
  3. **Contrasting traits:** The alternative forms of a character are called **contrasting traits.**
  4. **Diploid**: Each species possesses a specific number of chromosomes in the cell. This chromosomal component in the cell is called **diploid number** (2n).
  5. **Haploid**: During the formation of gametes, diploid number of chromosomes is reduced to half during meiosis. This chromosomal component in the gametes is called **haploid number (n).**
  6. **Homologous chromosomes:** Chromosomes which occur in pairs in diploid cells and are identical in size, structure and behaviour. In a homologous pair, one chromosome is *"maternal"* (comes from maternal parent) and the other is *"paternal"* (comes from paternal parent).
  7. **Homozygous:** When the maternal and paternal chromosomes of a homologous pair carry identical alleles at the same locus (TT or tt), the genotype is described as **homozygous** and the organism is a **homozygote**.
  8. **Heterozygous:** When the maternal and paternal chromosomes carry different alleles at the same locus, the karyotype is called **heterozygous** and the organism is heterozygote.
  9. **Gene:** A gene is the fundamental physical unit of heredity.
  10. **Gene locus:** Gene occupies a specific position called locus on a chromosome. A chromosome is composed of a large number of linearly ordered units called genes (Mendel's unit factors). The location on a given chromosome where any particular gene occurs is called gene **locus** (pl. loci).
  11. **Alleles:** The two alternative forms of a gene are called **alleles**. One of the alternative forms of a gene that occurs at a given locus in a chromosome is the **allele.** For example the allele responsible for tallness is T' and the allele responsible for dwarfness is 't'.

THE MONOHYBRID CROSS :

- **Monohybrid cross is the cross involving only a pair of contrasting traits of a single character.**
- It is conducted by crossing individuals from two parent strains (pure lines).
- Each parent exhibits one of the two contrasting traits of a character.
- In the pea plant **Tall** and **dwarf** represent contrasting traits of the character **height of the stem.** These tall and dwarf are pure lines.
- Mendel crossed a homozygous tall (TT) plant to a dwarf (tt) plant. This generation is called **P_1,** or **parental generation.**
- The offspring produced on crossing the two pure line parents of contrasting traits represents the **F_1,** or **first filial generation.**

- The F_1 generation, on crossing a homozygous tall plant to a dwarf plant, consisted of hybrid tall plants (Tt).
- Mendel self pollinated the F_1 generation tall plants to produce F_2 **generation, or second filial generation.**
- Mendel observed that 787 of 1064 F_2 plants were tall, while 277 of 1064 were dwarf. The ratio of tall to dwarf i.e. **phenotypic ratio** is about 3 : 1 in the F_2 generation.
- The **genotypic ratio** of the F_2 generation is 1 : 2 : 1 (1 = TT, 2 = Tt, 1 = tt).
- The genotypes and phenotypes resulting from the fertilization of gametes can be easily represented by constructing a **punnet square,** named after the discoverer **Reginold. C. Punnet.**
- Mendel called tall trait, a **dominant** as it appeared in the heterozygotes or hybrids of F_1 generation.
- He also observed that dwarf trait disappeared in F_1 generation and reappeared in the F_2 generation.
- So called it as **recessive.** Mendel continued the experiment upto F_3 generation to find out whether all the tall plants of F_2 generation are similar.
- The F_2 plants were self pollinated to produce the F_3 generation.
- Some of the tall plants produced the tall plants only and the other tall plants produced the tall and dwarf plants on self pollination.
- So the tall plants in the F_2 generation are not similar genotypically. Dwarf plants produced only dwarf plants.
- Mendel conducted similar experiments with the other characters of *Pisum sativum.*
- Results were found to be similar with all the seven characters.
- He conducted crosses in two different ways (reciprocal cross) for each character. That is pollen from the tall plant pollinating with dwarf plant and pollen from dwarf plant pollinating with tall plant.
- **The cross between different parents with sexes reversed is known as reciprocal cross.**
- Results were found to be same in either way of the cross.

Mendel's first three postulates :

- Using the consistent patterns of results of the monohybrid crosses, Mendel derived the following three postulates of inheritance.

1. Unit factor in pairs:

- Hereditary character is controlled by unit factor that exists in pairs in individual organisms.
- A specific unit factor exists for the trait **tall (T)** and another for the trait **dwarf (t).**
- As these factors occur in pairs, three combinations are possible; two factors for tall (TT), two factors for dwarf (tt) or one factor for each trait (Tt).
- These factors serve as basic units of heredity and are passed from generation to generation.

2. Dominance/recessive:

- In a pair of contrasting traits of a character, one is dominant trait and the other is the recessive trait.
- The trait that is expressed in the F_1 generation is called as **dominant trait** and the factor for this trait is **dominant factor** (tall is the dominant trait, the dominant factor is 'T').
- The other trait which is not expressed is called as **recessive trait** and the factor is **recessive factor** (dwarf is the recessive trait and the recessive factor is 't').
- When individual is heterozygous with two unlike unit factors of a character (Tt), one unit factor is dominant (T) to the other and the other is recessive (t).
- A dominant trait will be expressed with even a single dominant unit factor, but a recessive trait will be expressed only with two recessive unit factors of that trait.

3. Segregation:

- This law states that **the two unit factors** segregate **during the gamete formation. These gametes are pure for the factors they** possess. **The paired condition is restored by the random fusion of gametes during fertilization.**
- During the formation of gametes due to meiosis the paired unit factors (alleles) separate or segregate randomly with equal probability so that each gamete receives only one of these factors in equal proportions.
- The pure dominant plant (pure TT) produces gametes with the dominant factor ('T') and the recessive plant (tt) produces gametes with the recessive factor ('t').
- If an individual contains dissimilar factors (Tt), two different types of gametes are produced. Each gamete contains either the allele 'T' or 't' in equal proportions.
- Each gamete has 50% probability of getting either dominant or recessive factor This segregation is considered to be the **first law of genetics or purity of gametes.**

Single factor test cross

- Tall plants produced in F_2 generation are either TT or Tt genotypes Mendel devised a simple method i.e. tcstcross as a supporting cross to distinguish between the heterozygous dominant and homozygous dominant genotypes.
- **A cross between an organism of dominant phenotype with an unknown genotype and a known recessive phenotype (homozygous) is called testcross.**
- For instance the organism of the dominant phenotype, (whose genotype is not known), is crossed to a recessive individual.
- Tall is crossed to a recessive dwarf plant. If the progeny is tall and dwarf in the ratio 1:1, the tall is **heterozygous (Tt).**
- If the progeny is all tall, the tall is **homozygous** (**TT**).
- **Crossing of F_1 hybrid to any of the P_1 parent (or an organism with genotype similar to the P_1 parents) is called backcross.**

THE DIHYBRID CROSS

- **The cross involving two pairs of pure line contrasting traits is called a dihybrid cross. Two factor cross is a genetic cross involving two characters.**
- The true breeding pea plant with **yellow cotyledons** and **round (YY RR) seeds** is crossed to a plant with **green cotyledons and wrinkled seeds (yy rr).**
- All the F_1 offspring are yellow round phenotypically and double heterozygous genotypically (Yy Rr).
- So round is dominant over wrinkled, yellow is dominant over green phenotypes.
- The F_1 individuals are self pollinated to produce the F_2 generation.
- Approximately 9/16 of the F_2 plants are yellow round, 3/16 are yellow wrinkled, 3/16 are green round and 1/16 is green wrinkled.
- The phenotypic ratio of F_2 generation is **9 : 3 : 3 : 1** and the genotypic ratio is **1 : 2 : 2 : 4 : 1 : 2:1:2: 1.**
- If only a pair of contrasting traits is considered the phenotypic ratio is 3 : 1; round and wrinkled is 12:4 = 3:1 and Yellow and green is 12 : 4 = 3 : 1. The results are shown in the Fig: 6.1.4.
- The F_2 progeny showed not only the parental combinations i.e. yellow round and green wrinkled phenotypes, but also the new combinations i.e. yellow wrinkled and green round phenotypes.
- These new combinations are produced due to genetic recombination by independent assortment.
- This recombination is called **Mendelian recombination.**
- Mendel also crossed a yellow wrinkled phenotype i.e a dominant and a recessive combination (YY rr) to a green round (yy RR) phenotype.
- The results of the F_1 and the F_2 generations were found to be similar, in both crosses.
- Based on the dihybrid cross Mendel proposed the concept of independent assortment.

Mendel's fourth postulate, the independent assortment of chromosomes

- When two or more pairs of contrasting traits are brought together in a cross, the inheritance of one pair of traits is independent of the other pair of traits.
- Law of independent assortment states that **random distribution of alleles to the gametes occurs when genes are located on different chromosomes.**
- **The distribution of one pair of alleles is independent of the other pair.**
- Theoretically a dihybrid cross consists of two monohybrid crosses. Thus random distribution of alleles to the gametes occurs when these genes are located on different homologous pairs.
- During gamete formation, allelic pairs or unit factors Yy, Rr segregate independent of each other. As a result of segregation a gamete receives one member of every allelic pair of unit factors ('YR', 'Yr', 'yR', 'yr').
- One factor does not influence the segregation of the other. All possible combinations of gametes will be formed in equal proportions (i.e. 'YR' : 'Yr' : 'yR' : 'yr' =1 : 1 : 1 : 1).
- *The independent assortment of alleles occurs due to the location of different allelic pairs on different homologous pairs.*
- This independent assortment is considered as **the second law of genetics.**

Two factor testcross

- **The cross between the individual with two heterozygous pairs of alleles with double recessive parent is two factor testcross.**
- It is conducted generally to know the genotype of the unknown.

- The yellow, round phenotype in the F_2 generation may result from YY RR, YY Rr, Yy RR and Yy Rr genotypes.
- If an F_2 yellow, round plant is crossed to the homozygous recessive green wrinkled plant, analysis of the offspring indicates the genotype of the yellow round plant.

If the plant of unknown genotype is heterozygous for both the traits, the four phenotypic combinations in the offspring appear in equal proportion.