Introduction to Genetics and Mendelism

Contents: Definitions**;** Pre-Mendelian era; A short life-sketch of Mendel**;** Rediscovery of Mendel's principles**;** Mendel's laws of inheritance**;** Contrasting characters that Mendel studied**;** Reasons for Mendel's success; Some common terminologies used in Genetics; Suggested reading.

Definitions

Genetics (Gr. *Gen*- to generate; to grow into) is the science of heredity and variation. **Heredity** (L. *hereditus*- heir ship) refers to relation between successive generations; *i.e.* similarities between parents and offspring or progenies. **Variation** (L. *variare*- to change) means dissimilarities that exist between parents and offspring.

Pre-Mendelian era

A brief note on pre-Mendelian era (1715-1860) presented below would be helpful in understanding the events for Mendel's discovery.

1715: First artificial hybridization in *Dianthus* by Thomas Fairchild

1761-66: Joseph G. Kolreuter studies hybridization in plants; he got some results on backcross and correctly identified the roles of father and mother in heredity

1778: William Herbert observed new varieties from hybridization experiments

1822: John Goss crossed varieties of garden peas and observed dominance and segregation phenomena, but he did not record any data

1823: Similar to John Goss, Thomas A. Knight also observed the segregation in plants and realized the importance of hybridization in the improvement of crops without preserving his experimental data

1816-1860: Lous de Vilmorin observed the segregation of flower colour in *Lupinus hirsutus*

Even though the above-mentioned scientists experimented, observed and realized various aspects of plant heredity, they did neither preserve nor record their data properly. As a result, they were unable to analyze, interprete and explain their findings.

A short life-sketch of Mendel (1822-1884): The father of Genetics

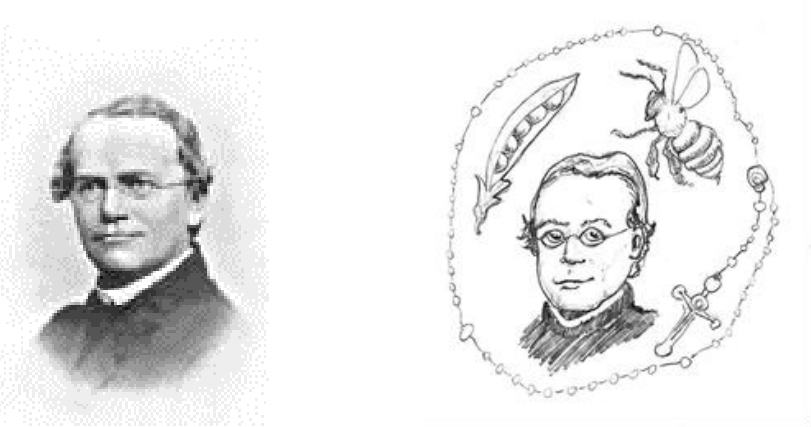

Fig 1.1 Portraits of Johann Mendel (1822-1884) depicting his profession and hobbies

Johann Mendel, son of Anton Mendel and Rosine Mendel; Elder sister Veronika Mendel, younger sister Theresia Mendel; they belong to a German speaking, peasant family.

1822- 22 July, born in Heinzendorf, Moravia, Austria (now a part of Czech Republic); started education in a village school, obtained record marks; Mendel was interested in gardening and beekeeping

1840- Studied at Olomouc University with hardship and graduated in Philosophy and Physics

1843- Joined as a monk (friar) in the Augustinian Monastery, Brünn, Austria

1847- Became priest and was titled 'Gregor'

1849- Appointed as a substitute teacher in a high school to teach Mathematics and Greek

1851- Went to the University of Vienna to study math, physics, natural science, physiology and palaeontology, but could not succeed

1854- Returned to Brünn and continued teaching up to 1868

1857-1865: Collected four species of garden peas (*P. sativum, P. quadratum, P. saccharatum, P. umbellatum*) belonging to 34 varieties and conducted his experiments in the monastery garden; tested some 28,000 plants in the 2 hector monastery garden

1865- Delivered two lectures on 8th February and 8th March on 'Experiments on Plant Hybridization' to Brünn Natural History Society

1866- Mendel's paper was published in Austrian language, but little attention was paid to his findings

1868- Retired from school job and became an Abbot; the last days were full of frustrations and conflicts between municipal and church authorities

1884- Died of *chronic nephritis* on the 6th January at the age of 61+ and was buried in Brünn Central Cemetery on the 9th January

Note: Perhaps Charles Darwin (1859) was unaware of Mendel's paper. Wilhelm von Waldeyer-Hartz (1888) discovered chromosomes 4 years after Mendel died.

Rediscovery of Mendel's principles and Establishment of Mendelism

1900- 16 years after Mendel's death, three scientists rediscovered Mendel's principles independently. They were: Dutch botanist, Hugo deVries; German botanist, Carl Correns; and Austrian botanist, Erich von Tschermak-Seysenegg.

1905-06- An English biologist Sir William Bateson coined the term 'Genetics' and he formulated the genetic principles from Mendel's findings; Bateson therefore is known as the 'Founder of Modern Genetics'

1909- Wilhelm Johannsen, Belgian botanist, coined the term 'gene' (took it from Darwin's pangene) for the hereditary unit 'factor' proposed by Mendel (1865). Johannsen also coined the terms 'genotype' and 'phenotype'

Mendel's (1865) four basic postulates:

1. Unit 'factors' exist in pairs and they are responsible for the inherited traits;
2. During the formation of gametes, the unit factors are separated so that each gamete gets only one factor of a given pair (*Law of segregation*);
3. When two factors for the alternative expression of a trait are brought together in fertilization, the resulting individual will show only the dominant trait (*Law of dominance*); and
4. When two or more pairs of factors are involved, they assort or combine at random during the formation of gametes (*Law of independent assortment*).

Note: Bateson (1906) translated Mendel's original paper in English, and formulated the genetic principles (Mendel's laws of inheritance) from Mendel's findings.

Mendel's laws of inheritance

Mendel's 1st Law: The law of segregation

During the formation of gametes, the unit 'factors' are separated so that each gamete receives only one 'factor' for a given pair.

Mendel's 2nd Law: The law of independent assortment

When two or more pairs of 'factors' are involved, they assort or combine at random during the formation of gametes. **Note:** There are exceptions to both laws of Mendel.

Contrasting characters that Mendel studied

After initial trials and errors, Mendel finally chose to work with seven pairs of contrasting characters in garden peas that are presented in the table below and in Fig 1.2.

Characters	Dominant	Recessive
1. Length of stem	Tall (T)	dwarf (t)
2. Forms of the ripe seeds	Round (R)	wrinkled (r)
3. Colour of seed cotyledons	Yellow (Y)	green (g)
4. Form of the ripe pods	Inflated (I)	constricted (i)
5. Colour of the seed coat	Grey (G)	white (g)
6. Colour of the unripe pods	Green (Gr)	yellow (gr)
7. Position of the flower	Axial (A)	terminal (a)

Note: Fortunately, the traits that Mendel chose for his experiments were located on seven different pairs of chromosomes in *P. sativum.*

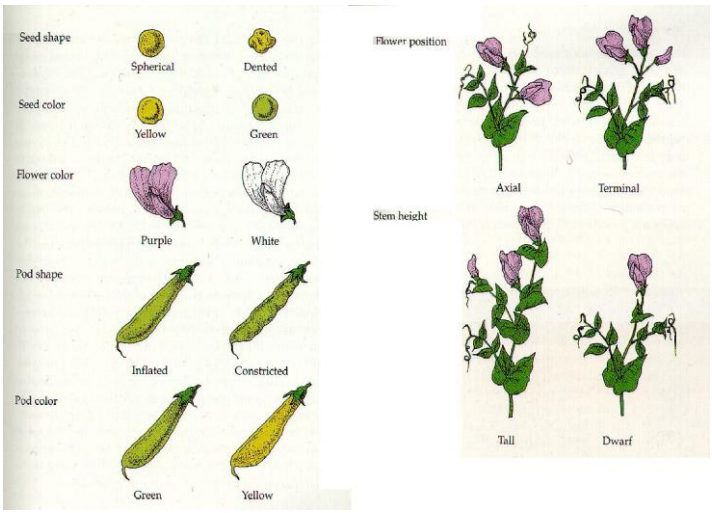

Fig 1.2 The contrasting characters that Mendel studied with his garden peas

Reasons for Mendel's success

1. The plants were easy to cultivate and ideal for controlled breeding;
2. From 34 varieties, Mendel selected 22, which all showed quite distinct and clear contrasting characteristics;
3. He spent some 2 years to ascertain these characteristic features;
4. The plants were self-pollinating, but artificial crossbreeding was possible, and the resulting hybrids were fertile;
5. Mendel worked on one trait at a time;
6. He kept a meticulous record of all his observations, used statistical methods and applied the law of probability;
7. He was fortunate enough in choosing the 7 pairs of characters and incidentally, the pea plant has 7 pairs of chromosomes;
8. The contrasting characters did not show any interactions and linkage.

Some common terminologies used in Genetics

Gene: A fragment of DNA molecule that specify for a particular amino acid or protein. It is a particulate determiner of hereditary trait and is located in a fixed place on the chromosome.

Genotype: The genetic makeup of an individual, usually expressed by a symbol, *e.g*. TT, Tt, tt etc.

Phenotype: The appearance of an individual, usually expressed in words, *e.g*. tall, dwarf, wild-type, taster, non-taster etc.

Homozygote (adj. homozygous): An individual whose chromosomes bear identical genes of a given allelic pair and so produce only one kind of gamete, *e.g*. TT, aa, DD etc.

Heterozygote (adj. heterozygous): An individual whose chromosomes bear unlike genes of a given allelic pair and so produce more than one kind of gamete, *e.g*. Tt, Aa, Dd etc.

Homologous chromosomes: Chromosomes occurring in pairs, derived from each of two parents, morphologically alike and bearing exactly the same gene loci.

Hybrid: An individual resulting from s cross between two genetically unlike parents, *e.g*. TT × tt → Tt. A hybrid is usually a heterozygote.

Locus (pl. loci): The position on a chromosome occupied by a particular gene or one of its allele, *e.g*. white eye (w) gene is located at 1.5 locus on X chromosome of *Drosophila melanogaster*.

Allele (Allelomorph): One member of a pair (or a series) of genes that occupies at a particular locus on homologous chromosomes.

Offspring (Progeny): Individuals derived from parents.

Monohybrid cross: A cross between two parents that differ in only one heritable character under consideration, *e.g*. TT × tt.

Dihybrid cross: A cross between individuals differing in two gene pairs under consideration, *e.g*. TTRR × ttrr.

Trihybrid cross: A cross between individuals differing in three gene pairs under consideration, *e.g*. TTRRYY × ttrryy.

Polyhybrid cross: A cross between individuals differing in more than three gene pairs under consideration, *e.g*. AABBCCDD × aabbccdd.

Back cross: The cross of a progeny individual with one of its parents, dominant or recessive, *e.g*. Tt × →TT or tt

Test cross: The cross of a progeny individual with its recessive parent, generally to determine whether the individual of dominant phenotype is homozygous or heterozygous, *e.g*. Tt × tt →TT: tt

Dominant: Member of a pair of alleles that expresses itself in heterozygote to the complete exclusion of the other member of the pair, *e.g*. T is dominant over t.

Recessive: Member of a pair of alleles that fails to express itself in the presence of its dominant allele; expresses only in the homozygote state, *e.g*. t is recessive to T.

P: Parental generation in a given cross, *e.g*. P: TT × tt (a cross between pure tall and pure dwarf).

F_1: The first filial generation, *e.g*. F_1: Tt (all tall plants in heterozygous state).

F_2: The second filial generation, *e.g*. F_1 selfing (Tt × Tt): ¼ TT: ½ Tt: ¼ tt.
Punnett square (checkerboard): A grid designed to determine all possible genotypes produced by a given cross as shown below (Fig. 1.3).

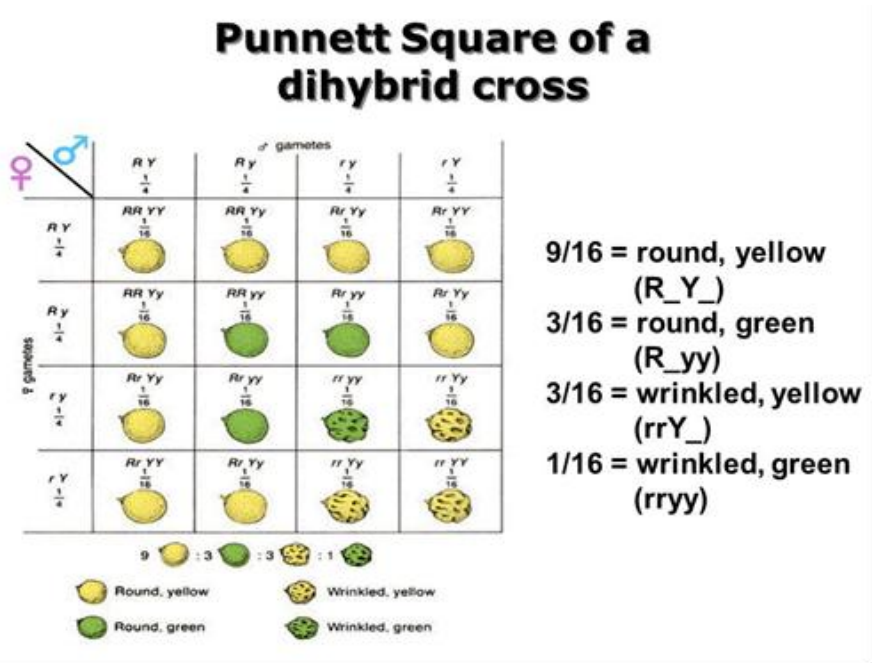

Fig 1.3 Punnett square for a dihybrid cross that resulted in 9: 3: 3: 1 ratio in F_2

Suggested reading:

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ইসলাম, ম.সা., খান, হা.সা. ও রানা, ম.হা.তা. ২০১৭। *জেনেটিক্স: মিল ও অমিলের বিজ্ঞান*। অন্যপ্রকাশ, ঢাকা।