Mendelian Inheritance

We have seen in our discussion of gametogenesis (sporogenesis that the gametes or the spores produced by the maturation proces essentially have the haploid chromosomes number. While the mal gamete (sperm) is mainly composed of chromosomes, the female gamete (egg) possesses cytoplasmic material in addition to the chromosomes. The union of these two gametes initiates a new individual, whose subsequent growth is guided by the genetic information contained in the chromosomes in the form of genes. A diploid (2n) organism has a double dose of a gene whereas a haploid (n) has a single dose. It is the genes arranged in a linear fashion on the chromosomes which are passed along with the chromosomes from generation to generation and determine the limits of growth of an individual. The chromosomes thus being the carriers of genes are the physical basis of heredity.

Brief life sketch of Mendel

Gregor Mendel (1822—1884), who is called the father of genetics, was born in a poor farmer family of Austria. After completing education upto secondary school, he joined a monastery during 1843, and on his 25th birthday he became a monk. Mendel conducted hybridization work on various plants, i.e., snapdragon, pumpkin, flax, bean, pea, plum, pear, maize, etc. He was sent to Vienna University during 1851 to study science and mathematics. On his return to Brunn (1854), in addition to his duties as priest, he was appointed a substitute teacher in a school. He collected garden pea seed from the market during 1857 and started regular hybridization work and was thus able to present his results before the History Society of Brunn in 1865. The results were published in 1866 and were sent to various countries. His work remained unnoticed for a period of 34 years. In 1900 three biologists, Correns, deVries, and Tschermak read Mendel's forgotten paper and showed that they had also obtained similar results from their own experiments.

Mendel's choice of pea plant was not a mere chance, but was based on these facts: (i) many pure-breeding pea varieties were available

(ii) floral structure and mode of pollination in pea ensures self-pollination and (iii) hybrids are fertile. He finally selected the following seven easily recognisable contrasting characters:

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No.	*Character*	*Dominant*
1.	Tall versus dwarf vine	Tall
2.	Green versus yellow colour of unripe pods	Green
3.	Inflated versus constricted pods	Inflated
4.	Axillary versus terminal flowers	Axillary (Axial)
5.	Yellow versus green endosperm (Cotyledon)	Yellow
6.	Round versus wrinkled seed surface	Round
7.	White versus grey seed coat	Grey

Mendel made several crosses of pea plants having contrasting characters to find out how the hereditary elements behaved while passing from parents to offspring. We shall consider here only one or two of the several experiments that he performed.

For instance, Mendel crossed a tall pea plant with a dwarf one and observed that all the resulting hybrid plants (F_1) were tall. The F_1 plants were self-fertilized to produce F_2 seed which was planted to study the behaviour of the F_2 generation. Mendel noted that of all the F_2 plants, 3/4 plants were tall and 1/4 plants were dwarf.

It may be pointed out here that the outward expression of a character later came to be referred to as "phenotype" and the complex of genetic factors controlling this expression as "genotype". We may now write the above cross genotypically using symbols as follows:

Parents × (Tall) TT, tt (Dwarf)
Gametes T, T : t, t,

Combination of female and male gametes produces F_1 generation. } F_1 Tt (Tall)

On selfing, two types of gametes (pollens and ovules) are possib from Tt plant and they are T and t. The random union of the male T an t with the female T and t will produce the F_2 generation as follows:

Male gametes T, t
Female gametes T, t

TT Tt
Tt tt

Genotypes in F_2 ITT 2Tt Itt
Phenotypes being 3 Tall: 1 Dwarf

The same combinations can be obtained by the checker-boar method as:

		male gametes $\frac{1}{2}$T	male gametes $\frac{1}{2}$t
female gametes	$\frac{1}{2}$T	$\frac{1}{4}$TT	$\frac{1}{4}$Tt
	$\frac{1}{2}$t	$\frac{1}{4}$Tt	$\frac{1}{4}$tt

F_2 phenotypic ratio: 3 tall : 1 dwarf or $\frac{3}{4}$ tall : $\frac{1}{4}$ dwarf
F_2 genotypic ratio: 1 tall (TT) : 2 tall (Tt) : 1 dwarf (tt)
or $\frac{1}{4}$ (TT) : $\frac{1}{2}$ (Tt) : $\frac{1}{4}$ (tt).

When Mendel selfed these F_2 plants, he observed that the dwarf plants, which were $\frac{1}{4}$ of the total F_2 population, produced all dwarfs, while out of the $\frac{3}{4}$ tall plants, $\frac{1}{3}$ produced all tall and $\frac{2}{3}$ produced both tall and dwarf plants: that is precisely what was expected from the genotypes contained in the F_2 population above.

Law of Segregation

All other 6 characters behaved in the similar manner in crosses. From this, Mendel formulated his first law of inheritance: *the law of Segregation or Mendel's First law of Inheritance.* It states that (1) hereditary characters are determined by some particulate factors (genes); (2) these factors occur in pairs and (3) at the time of gamete formation segregate at random so that only one member of a gene pair is transmitted to a particular gamete.

Dominance

The other important phenomenon which Mendel discovered was that of *dominance*. He noticed that the hybrids (F_1's) between tall and dwarf were all tall like the tall parent, although it also inherited the dwarf factor. The factor which dominated the effect of the other allele in the F_1 was designated by Mendel as dominant and the alternative factor that remained latent in F_1 as recessive. He showed the dominant characters by the use of capital letters and the recessive by small letters while explaining his results: –

> *Note:* In crosses, it is not essential for one character to behave always as dominant, since situations midway between the two parents are also obtained in F_1.

Mendel's law of segregation and its explanation

Mendel's first law of inheritance is the law of segregation, which explains the behaviour of alleles of a gene pair in a diploid organism at gamete formation. According to this law, the two alleles which may be identical or dissimilar i.e., AA, aa or Aa, segregate by going to different poles in meiotic divisions and thus end up into different gametes. Note that segregation of alleles occurs during the meiotic divisions.

Consider a cross between two true-breeding plants. One of them is tall and the other dwarf. Because both of them are diploid, each has a pair of alleles. The tall one has both the alleles, T, T and the dwarf one has both its alleles, t, t. The gametes which they will produce will have one allele only as the number is reduced to one half. We shall get a gamete of T constitution from one parent and a gamete of t constitution from the other. They reunite and the act of their reunion is called fertilization. The new plant produced by the union of T and t will be Tt. This means that the new hybrid plant has two alleles but they are not alike, as one of them is T (for tallness) and the other is t (for dwarfness). If the hybrid plant is tall, we will be able to say that the character tallness controlled by T is dominant over the corresponding contrasting character, dwarfness, controlled by t. Further, when this hybrid plant Tt matures and forms gametes, the two alleles T and t, carried on the two homologous chromosomes, cannot stay together, because the homologous chromosomes must go to the opposite poles in meiotic division and so will the two contrasting alleles. This going of the two alleles to the opposite poles in meiosis provides the mechanism for segregation of the alleles of the same gene pairs. Consequently, two types of gametes, T and t are formed by the hybrid plant in its male and female reproductive organs. Their reunion will produce these offsprings: 1 TT : 2 Tt : 1 tt, as explained earlier.

From this discussion, it is apparent that the two alleles remain together in a diploid individual and segregate when that individual forms gametes and again come together in various combinations when male and female gametes unite to produce new individuals.

Mendel's second law of inheritance or Mendel's law of independent assortment

We may now consider two characters together as to their behaviour in inheritance. For instance, Mendel crossed a tall plant having axillary flowers with a dwarf plant having terminal flower. Character dominance is indicated on page 18. As before, T and t represent tallness and dwarfness, respectively, and let A and a represent axillary and terminal flower positions, respectively.

We may write the cross genotypically as follows:

Parents TTAA (tall axillary), ttaa (dwarf terminal)
Gametes TA ta
F_1 TtAa (tall with axillary flowers)

The F_1 plants will produce these four types of gametes TA, Ta, tA, ta, in equal proportion. The four types of gametes will be male as well as female and their random union will produce 16 combinations in the next generation (F_2).

Female gametes 1/4 TA + 1/4 Ta + 1/4 tA + 1/4 ta
Male gametes 1/4 TA + 1/4 Ta + 1/4 tA + 1/4 ta

1/16 TTAA + 1/16 TTAa + 1/16 TtAA + 1/16 TtAa
1/16 TTAa + 1/16 TTaa + 1/16 TtAa + 1/16 Ttaa
1/16 TtAA + 1/16 TtAa + 1/16 ttAA + 1/16 ttAa
1/16 TtAa + 1/16 Ttaa + 1/16 ttAa + 1/16 ttaa

These sixteen combinations for the dihybrid cross can also be calculated by the checker-board method:

F_1 TtAa

Gametes TA, Ta, tA, ta

		Male gametes			
		TA	Ta	tA	ta
Female gametes	TA	TTAA	TTAa	TtAA	TtAa
	Ta	TTAa	TTaa	TtAa	Ttaa
	tA	TtAA	TtAa	ttAA	ttAa
	ta	TtAa	Ttaa	ttAa	ttaa

Mendel noted that all F_1's were tall with axillary flowers where from he confirmed that tall was dominant over dwarf and axillary flower over terminal flower, and that out of these 16 combinations of the F_2, 9 were tall with axillary flowers, 3 were tall with terminal flowers, 3 were dwarf with axillary flowers and only one was dwarf with terminal flowers. It may be seen that, as the 9 tall, axillary-flowered plants have at least one T and one A, these show both the dominant characters, 3 have T but no A, they show one dominant character, the other 3 have A but no T, and they show the other dominant character, one has neither T nor A but is ttaa and is double recessive.

Explanation. It will be noticed that when the hybrid plant, TtAa, matures, it forms four types of gametes (pollens as well as ovules). T and t are the two members of an allelic pair and gametes are formed as a result of meiotic divisions, whereby the two alleles must segregate. In the case of the hybrid plant TtAa, T and t must segregate, i.e., they must not be present in the same gamete and similarly, A and a must segregate and go to different gametes. But while the two members of a given allelic pair must segregate, they do so independently of the members of the other allelic pair, that is, if T goes to one pole, either A or a of the allelic pair Aa, can go with T to the same pole. So the resulting gamete (pollen or ovule) can have T with A (TA) or T with a (Ta). Similarly, the gamete produced at the other pole can have t with A (tA) or t with a (ta).

Accordingly, this plant, TtAa, must produce four types of gametes, i.e., (TA), (Ta), (tA) and (ta) in equal proportion. This is actually what the plant does.

The production of these four types of gametes in equal numbers is possible only if the two gene pairs, Tt and Aa assort independently of each other. This phenomenon is called "independent assortment" of characters, the Second Mendelian Law. If the four types of gametes, male as well as female, unite at random in the process of fertilization, 16 F_2 combinations as shown above should be possible.

"The segregating genes assort independently."

Back-cross and Test cross

When F_1 hybrid is crossed with one of its parents, it is called a backcross; when the cross involves the recessive parent, it is called a test cross. We may remember that the F_1 hybrid between Mendel's tall and dwarf pea plants were all tall, which may be crossed with dwarf recessive (tt) parent to form a test cross. The hybrid tall parent (Tt) would produce equal number of *T* and *t* gametes and the dwarf being homozygous should form only one type of gametes, i.e., t. Half the ovules of the genotype, *T* when fertilized by *t* pollen will produce *Tt* plants and the other half ovules of *t* genotype fertilized by *t* pollen produce *tt* plants. A definite ratio of 1/2 tall and 1/2 dwarf plants would thus be obtained. The 1:1 ratio based on theoretical calculations was confirmed by Mendel through such experiments. Mendel clearly demonstrated that the characters are determined by particulate factors which do not blend with, or contaminate, one another during the course of their transmission from generation to generation.

Similarly, results from a dihybrid situation were also tested by Mendel. He crossed yellow round with green wrinkled plants to be symbolized genotypically as YYRR & yyrr, respectively. The F_1 hybrid would be YyRr (yellow-round), which when backcrossed to double recessive yyrr parent will result in four phenotypes in equal number, i.e., *Yellow-round* YyRr; *yellow wrinkled* Yyrr; *green round* yyRr; and *green wrinkled* yyrr (Dihybrid test cross ratio 1:1:1:1).

This proved that:

1. The factors responsible for character expression are particulate in nature.
2. Segregation of factors takes place which shows that they do not blend with one another.
3. Grouping or the assortment of the factors is random during the process of gamete formation.
4. 1/4 : 1/4 : 1/4 : 1/4 test cross ratio proves that all types of gametes were produced in equal number and that they had an equal chance for union amongst themselves.

Blending theory

Before Mendel's work on hybridization was duly recognised, the "Blending theory" enjoyed popular support as the basis of inheritance of characters in living organisms. According to this theory, "heredity determinants were fluid-like which would always produce a blended expression of the characters of the two mating individuals." For instance, if one parent has black body colour and the other white, their offspring must all be grey, which cannot be separated again in black and white in the progeny.

Particulate theory

Mendel contradicted the above idea by showing from his experimental evidence that the determiners of heredity are individual particles, which, in various combinations, can produce numerous shades of expression of a character and that these determinants are separable unaltered at the time the individual forms gametes. This theory is called the particulate theory of inheritance and is now accepted as valid.

Resume of Mendel's Experiments on Pea

Mendel made experiments on pea using seven different contrasting characters. He selected pea plant as his experimental material because of the following advantages that the pea plant offered him:

1. Crossing was easy.
2. The characters were easily distinguishable.
3. The hybrids were fertile and easily mutually crossable.
4. Due to special floral structure, contamination from foreign source was avoided.
5. Their culture was easy.

From his extensive data Mendel developed the following formula, which illustrate how fast the hybrid nature of a population disappears.

Segregating generation	**Genotype**			**Ratios**		
	AA	Aa	aa	AA	Aa	aa
1.	1	2	1	1 :	2 :	1
2.	6	4	6	3 :	2 :	3
3.	28	8	28	7 :	2 :	7
4.	120	16	120	15 :	2 :	15
n.	—	—	—	2^n-1 :	2 :	2^n-1

(*The Formula* $2^n-1 : 2 : 2^n-1$.)

$2^n-1 : 2 : 2^n-1$

2^{n-1} 2^{2-1} $4-1=3$

Conclusions:

$2^2-1 : 2 : 2^2-1$

3 : 2 : 3

1. Characters are controlled by heredity determiners (Genes).
2. These determiners (genes) can be dominant or recessive.
3. The genes segregate at the time of gamete formation (They do not get blended).
4. All possible combinations of genes occur in the gametes (Independent assortment).

5. The male and female gametes combine at random to produce offsprings showing all possible combinations of characters.

Reasons for Mendel's Success

1. His choice of the pea plant was excellent: ① the plant was self-pollinated and, therefore, highly homozygous, was ② easy to culture and to cross; ③ contamination of the crosses from the foreign pollen was avoidable due to its special floral structure.

2. Mendel started with simple experiments involving only one character difference and then proceeded with more complicated ones involving more than one character, making careful analysis of his data and testing them against his theoretical expectations.

3. He made use of mathematics to explain the biological behaviour of the factors determining character expression.

4. He was lucky that the characters he studied were not linked, otherwise he might have had difficulty in explaining the character variation not conforming with his hypotheses, as he had no knowledge of genes being located on the chromosomes.

Reasons for slow acceptance of Mendel's work

1. Some well-known scientists would have no confidence in Mendel's interpretation of the basis of inheritance, since they did not think Mendel was qualified enough to do that.

2. The results of some of the other scientists (Nagli's work on *hieracium*) were not in conformity with those of Mendel.

3. Mendel used mathematics to explain his results, whereas other scientists did not use this type of approach in the study of biological material.

4. Mendel was a kind of genius and a little ahead of his time. His contemporaries were ill equipped to understand him.

Intermediate dominance

In our previous discussion on Mendelian inheritance, we observed that in the F_1, one form of the character was dominant over the other. During later investigations, this generalisation did not hold good; hybrids showing expression intermediate between the two parents were observed. Where dominance exists, a single dose of the dominant gene produces the same effect as a double dose, but in case the dominance is lacking or incomplete the two alleles in the hybrid conditions (F_1) interact to produce expression more or less intermediate between the two original forms of the character. In a cross between red-flowered (AA) and white flowered (aa) snapdragons, the flower colour of F_1 (Aa) is pink, i.e., intermediate between red and white. This seems a sort of blending between the original colours. When the F_2 was grown, segregation occurred in the usual proportion of 1/4 red AA; 1/2 pink Aa; 1/4 white aa, which is actually a modification of the monohybrid phenotypic ratio (3:1). This shows that gene (A) alone is not sufficient to produce red flower colour, but the intensity of the red colour is reduced to pink when allelic constitution is (Aa). The segregation of the original colours showed that the heredity material did not blend in F_1. A backcross between the red AA and pink Aa produced 1/2 red and 1/2 pink and the backcross between white (aa) and pink (Aa) will produce 1/2 pink and 1/2 white. This shows that the characters are controlled by the particulate substance (genes) which segregate from one another uncontaminated.

Take another example: Some varieties of snapdragons have broad leaves; others have narrow leaves. In a cross between broad (BB) and narrow (bb) leaved varieties the F_1 (Bb) has intermediate leaves. In F_2 a segregation of 1 broad (BB) : 2 intermediate (Bb) and 1 narrow (bb) leaved plants is obtained. When flower colour and leaf shape were studied together i.e., the variety with red flower and broad leaves AABB was crossed with the white flower and the narrow leaf variety aabb; the F_1 had pink flowers and intermediate leaves. The F_2 segregated into 9 distinct phenotypes as follows:

1 AABB red broad : 2 AaBB pink broad : 2 AABb red intermediate : 4 AaBb pink intermediate : 1 AAbb red narrow : 2 Aabb pink narrow : 1 aaBB white broad : 2 aaBb white intermediate : 1 aabb white narrow. Each phenotype corresponds to a genotype. Here 9:3:3:1 ratio is modified to 1:2:1:2:4:2:1:2:1.

Physical basis of Mendelian inheritance

Mendel had no knowledge of the existence of chromosomes. The rediscovery of Mendel's work, attributing inheritance of characters to particulate factors (now called genes) inspired scientists to explain the nature of these factors. They argued that in order to be heredity determiners these particulate factors must possess the following attributes:

1. They should be continuous from one cell division to another.
2. They should be present in pairs in diploid organisms and separate out into different gametes at the time of gamete formation (segregation).
3. Each should have its own individuality and differ qualitatively from the other.
4. They should be capable of random reunion at the fertilization process.

In 1902 Sutton and Boveri put forth the *chromosomes theory of heredity* which correctly answered the above questions. According to the Sutton-Boveri hypothesis, genes (determiners) are present on the chromosomes in a linear order. 2 The chromosomes exist in pairs in the diploid organism (2n) and in the haploid organism the chromosome number is reduced to one half the original number (n) (Mendel's law of segregation). 3 Members of homologous pairs of chromosomes separate or segregate from each other during meiosis; their diploid number is restored at fertilization. 5 Two chromosomes carrying different genes assort independently of their original mates (homologues) depending upon the way they reorient themselves on the metaphase or equatorial plate (Mendel's law of independent assortment). 6 It is just a chance that the paternal and the maternal sets may go in original combination or may form new combinations.

It will be noted that the behaviour of the chromosomes conforms to that of the Mendelian factors (genes) and are, therefore, the physical basis of heredity. There is a close parallelism between the two things, genes and the chromosomes. To emphasise this fact once again, it may be repeated that:

1. Genes exist in pairs, because chromosomes, which carry them, exist in pairs.
2. Genes segregate at meiosis, because of the fact that chromosomes segregate.
3. Genes assort at random, because of the fact that chromosomes assort in this fashion.
4. Genes recombine during fertilization process in all possible combinations because chromosomes do so.

Some useful formulae

1. Number of kinds of secondary meiocytes $= 2^n$
2. Number of kinds of gametes2 $= 2^n$
3. Number of kinds of genotypes $= 3^n$
4. Number of kinds of phenotypes
   - (i) When dominance is incomplete $= 3^n$
   - (ii) When dominance is complete $= 2^n$
5. Number of kinds of homozygotes $= 2^n$
6. Number of kinds of heterozygotes $= 3^n - 2^n$
7. Number of kinds of gametic combinations 4^n

Where n—Number of heterozygous gene pairs.

PROBLEMS

1. What are the two most important laws of genetics that you have studied so far? Who formulated them and why did he succeed in enunciating these laws while others more qualified than him did not?