Genetics

Introduction.

The term genetics was coined by *Bateson* (1960). Genetics is the study of principles and mechanism of heredity and variations. The resemblance amongst offspring is never 100% (except in monozygotic twins) due to reshuffling of chromosomes and their genes. Same genetic traits are present in monoparental individuals formed through asexual reproduction or mitosis. Such individuals are called *ramets* while the whole group of similar individuals is called *clone*.

Father of genetics (classical genetics)	*Mendel*
Father of modern genetics/Animal genetics	*Bateson*
Father of experimental genetics/Drosophila genetics	*Morgan*
Father of human & physiological genetics	*Garrod*
Father of quantitative inheritance	*Kolreuter*
Father of Neurospora genetics	*Dodge*
Father of Eugenics	*Francis Galton*
Father of Indian genetics	*M. S. Swaminathan*

Heredity.

Heredity is the study of transmission of characters and variations from one generation to the next.

(1) **Basis of heredity :** Heredity involves the transfer of chromosomes from parents to offspring or one individual to another. Therefore, chromosome is the base of heredity. The physical basis of heredity are genes while chemical basis of heredity is DNA.

(2) **Pre-Mendelian view points**

(i) **Vapour and fluid theory :** Greek philosopher, Pythagoras proposed that some moist vapour is given out from the brain, nerves and all other parts of the body during coitus. On account of these vapours, the offspring exhibits similarities with the male parents.

(ii) **Semen theory :** Empedocles, suggested that both parents produce semen which arises directly from their various body parts. The semen from both the parents gets mixed and produces a new individual.

(iii) **Preformation theory** : Antony von Leeuwenhoek was the first to observe human sperms. This theory believes that one of the sex cells or gametes either sperm or egg, contained within itself the entire organism in perfect miniature form. Miniature form was called as 'homunculus'. The theory was supported by Malpighi, Hartosoeker and Roux.

(iv) **Particulate theory :** Maupertuis proposed that the body of each parent gives rise to minute particles. These particles unite together to form the daughter individual.

(v) **Encasement theory :** Charles Bonnet and his supporters presumed that every female contains within her body miniature prototypes of all the creatures which would descend from her, one generation within the other, somewhat like a series of chines boxes. This was named as encasement theory.

(vi) **Theory of epigenesis :** Wolgg proposed that the germ cells contain definite but undifferentiated substances, which after fertilization, become organised into various complex body organs that form the adult. This idea was referred to as epigenesis.

(vii) **Pangenesis theory :** This theory was proposed by *Charles Darwin* according to this theory every cell, tissue and organ of animal body produces minute invisible bodies, called gemmules or pangenes. They can produce offsprings.

(viii) **Weismann theory of germplasm :** August Weismann (1889) suggested the theory of continuity of germplasm. He described reproductive cells as germplasm and rest of the body as somatoplasm. The germplasm forms the bridge of life between successive generations and is passed on from one generation to the next.

(3) **Evidences against blending theory :** Thus individual would represent the mixture of both the parents. The prevailing view of in pre-mendelian era was blending theory. The hereditary material was thought of as being analogous to a fluid. Under this concept, the progeny of a black and white animal would be uniformly grey. The further progeny from crossing the hybrids among themselves would be grey, for the black and white hereditary material, once blanded, could never be seperated again. Pattern of inheritance shown by atavism also speaks against blending theory. The traits of sex do not blend in unisexual organisms.

(4) **Basic features of inheritance :** In the middle of 18th century, *Carolus Linnaeous* a Swedish taxonomist and two German plant breeders *Kolreuter* and *Gaertner* performed artificial cross pollination in plants and obtained hybrid offspring. Kolreuter obtained experimental evidence that inherited traits tended to remain discrete, although his observations were similar to mendel but he was not able to interpret them correctly. Mendel's great contribution was to replace the blending theory with particulate theory. Few essential features of inheritance are : –

(i) Traits have two alternative forms.

(ii) Traits are represented in the individual by distinct particles which do not blend or change.

(iii) Traits may remain unexpected for one or more generations and reappear later unchanged.

(iv) Traits may remain together in one generation and separate in a later generation.

(v) One alternative of a trait may express more often then the other.

Variations.

Variations are differences found in morphological, physiological and cytological behaviouristic traits of individuals belonging to same species race and family. They appear in offspring or siblings due to : –

- Reshuffling of genes/chromosomes by chance separation of chromosomes
- Crossing over
- Chance combination of chromosomes during meiosis and fertilization.

Types of variations

(1) **Somatic variations :** These variations influence the somatic or body cells. They appear after birth and are, also called *acquired characters, modifications or acquired variations*. Somatic variations are non-inheritable and usually disappear with the death of the individual. They are formed due to three reasons *i.e.* environmental factors, use and disuse of organs, and conscious efforts.

(i) **Environmental factors :** They have lesser effect on animals as compared to plants. Important environmental factors are as follows:

(a) **Medium :** Amphibious or emergent aquatic plants possess heterophylly, *i.e.* different types of submerged, floating and emerged leaves, e.g. *Ranunculus aquatilis*, *Limnophila heterophylla* and this meristic activities are due to change in depth and medium of water.

(b) **Light :** Partial shade causes elongation of internodes.

(c) **Temperature :** Plants of hot areas have extensive roots but smaller shoots. Human skin becomes darker with increase in environmental temperature.

(d) **Nutrition :** Honey bee larva feeding on royal jelly develops into queen while the ones obtaining ordinary nourishment (bee bread) grow into workers.

(e) **Water :** Water deficiency leads to several modifications in plants like succulente, spines, reduced leaves, thick bark, hair etc.

(ii) **Use and disuse of organs :** In higher animals and human beings, greater use of an organ leads to its better development as compared to other organs which are less used, e.g., stronger muscular body in a wrestler.

(iii) **Conscious efforts :** Acquired variations due to conscious efforts *include education*, training of pets boring of pinna, bonsai, etc.

(2) **Germinal variations :** They are inheritable variations formed mostly in germinal cells which are either already present in the ancestors or develop a new due to *mutations*. Germinal variations are of two types, continuous and discontinuous

(i) **Continuous variations :** They are *fluctuating variations* and also called *recombinations* because they are formed due to recombination of alleles as found in sexual reproduction. Darwin (1859) based his theory of evolution on continuous variations.

(ii) **Discontinuous variations :** They are *mutations,* which are ultimate source of organic variations. Discontinuous variations are caused by chromosomal aberrations, change in chromosome number and gene mutations. In pea seed coat colour changes gray to white is an example of spontaneous mutation.

Importance of variations

(1) Variations continue to pile up forming new species with time.

(2) They are essential in the struggle for existence.

(3) Adaptability is due to variations.

(4) Variations allow breeders to improve races of plants and animals.

(5) Discontinuous variations introduce new traits.

(6) Inbreeding between closely related organisms reduces variation.

Important terms used in inheritance studies.

(1) **Gene (Mendel called them factor) :** In modern sense an *inherited factor* that determines a biological character of an organism is called gene (functional unit of hereditary material).

(2) **Allelomorphs or alleles :** Alleles, the abbreviated form of term allelomorphs (meaning one form or the other) indicates alternative forms of the same gene. e.g., Tall **TT** and dwarf **tt** are alternation forms of the same gene etc.

(3) **Gene locus :** It is the portion or region on chromosome representing a single gene. The alleles of a gene are present on the same gene locus on the homologous chromosomes.

(4) **Wild and mutant alleles :** An original allele, dominant in expression and wide spread in the population is called wild allele. An allele formed by a mutation in the wild allele, recessive in expression and less common in the population is termed as mutant allele.

(5) **Homozygous :** Both the genes of a character are identical is said to be homozygous or genetically pure for that character. It gives rise to offspring having the same character on self-breeding e.g. **TT** (Homozygous dominant) or **tt** (Homozygous recessive).

(6) **Heterozygous :** Both the genes of a character are unlike is said to be heterozygous or *hybrid.* Such organisms do not breed true on self fertilization e.g. **Tt**

If we know the number of heterozygous pairs we can predict the following:

Number of types of gametes = 2^n

Number of $\mathbf{F_2}$ phenotype = 2^n (Where ***n*** is the number of heterozygous pairs).

Number of $\mathbf{F_2}$ genotype = 3^n

(7) **Genotype :** The genotype is the genetic constitution of an organism. **TT**, **Tt** and **tt** are the genotypes of the organism with reference to these particular pairs of alleles.

(8) **Phenotype :** Expresses the characters of individuals like form, sex, colour and behaviour etc.

(9) **Pure line :** Generations of homozygous individuals which produce offsprings of only one type *i.e.* they breed true for their phenotype and genotype.

(10) **Monohybrid, dihybrid and polyhybrid :** When only one allelic pair is considered in cross breeding, it is called monohybrid cross. Similarly when two allelic pairs are used for crossing, it is called dihybrid cross and more than two allelic pairs in a cross are called polyhybrid cross.

(11) **Reciprocal cross :** The reciprocal crosses involve two crosses concerning the same characteristics, but with reversed sexes.

(12) **Genome :** Total set of genes (DNA instructions) in the haploid set of chromosomes and inherited as unit from parents to offspring is called genome.

(13) **Gene pool :** All the genotypes of all organisms in a population form the gene pool.

(14) **F_1 Generation :** F_1 or first filial generation is the generation of hybrids produced from a cross between the genetically different individuals called parents.

(15) **F_2 Generation (Bateson, 1905) :** F_2 or second filial generation is the generation of individuals which arises as a result of inbreeding or interbreeding amongst individuals of F_1 generation.

(16) **Punnet square :** It is a checker-board used to show the result of a cross between two organisms, it was devised by geneticist, R.C. Punnet (1927). It depicts both genotypes and phenotypes of the progeny.

(17) **Back cross :** It is cross which is performed between hybrid and one of its parents. In plant breeding, back cross is performed a few times in order to increase the traits of that parent.

(18) **Test cross :** It is a cross to know whether an individual is homozygous or heterozygous for dominant character. The individual is crossed with recessive parent. The ratio will be 50% dominant and 50% recessive in

case of hybrid or heterozygous individual. In case of double heterozygote (e.g., RrYy) crossed with recessive (rryy) the ratio will be 1:1:1:1 test cross help to find out genotype of parents.

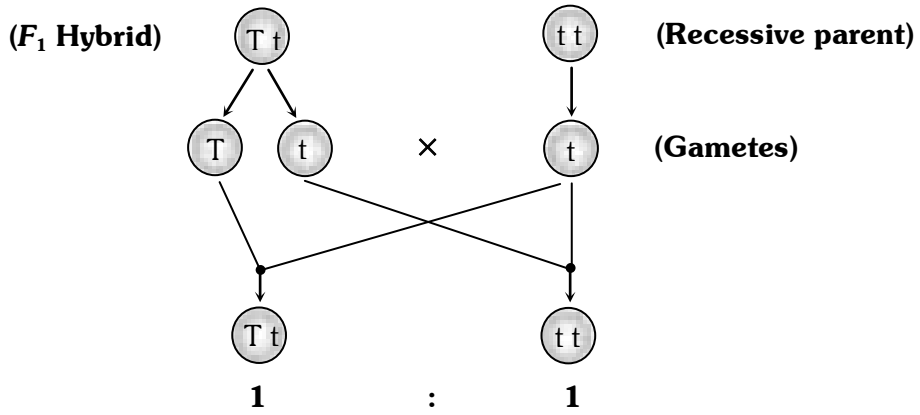


Fig : Showing test cross

(19) **Self cross/selfing :** It is the process of fertilization with pollen or male gametes of the same individual.

(20) **Theory of probability :** (i) Out of the two alternate events, the probability of occurrence of each one of them is 50%.

(ii) Two events are independent if occurrence of one does not affect the probability of occurrences of the other.

(iii) The probability of joint occurrence of two independent events is the product of their individual probabilities.

(iv) For an event, which can happen through two independent pathways, the probability of its occurrence is the sum of separate probabilities.

(21) **Observed Vs expected results :** Experimental results conform to the ones expected through the theory of probability if the size of the sample is small but they tend to approach the latter if the sample size is large.

(22) **Hybrid :** The organism produced after crossing of two genetically different individuals is called hybrid.

(23) **Heredity and variations in sexual and asexual reproduction**

(i) **Sexual reproduction :** Variations are common in animals and plants which reproduce by sexual means. The reason for this is that the sexual reproduction is biparental, involves meiosis and fertilization, and the offspring receives some traits from father and some from mother.

(ii) **Asexual reproduction :** Those organisms which reproduce by asexual means *eq*. bacteria, amoeba, euglena, rose etc. The asexual reproduction is monoparental, involves mitosis and the organism produced by it, inherits all the traits of its single parent. With the result, it is almost a carbon copy of the parent and is known as ramet. A group of ramets is called a clone.

Differences between clone and offsprings

Characters	Clone	Offspring
Type of reproduction	Clone is the product of asexual reproduction	Offspring is the product of sexual reproduction
Number of parents	Clone is monoparental	Offsprings is derived from two parents thus biparental

Cell division	Clone is formed by mitosis. meiosis does not occur.	Meiosis takes place prior to formation of gametes
Resemblance	Clone exactly resembles the parent	Offspring differs from parents.

Mendel's predecessors.

A number of scientists had worked on plant hybridization during the 18th and 19th centuries prior to the mendel. Some of the more notable scientists among them are *Joseph Koelreuter, John Goss, Gaertner, Darwin, Herbert,* and *Naudin Koelreuter* conducted extensive studies on hydridization between various species of Nicotiana (Tabacco) between 1761 and 1767, he noted the uniformity and heterosis in $\mathbf{F_1}$ (First ficial generation) and appearance of increased variations in $\mathbf{F_2}$. Koelreuter also observed that the hybrids were intermediate between their parents and that hybrids from reciprocal crosses were indistinguishable. Knight and goss conducted experiments on edible pea (*Pisum sativum*) much before Mendel but failed to formulate the laws of inheritance.

Mendelian period.

Introduction : *Gregor Johann Mendel* (1822-1884) first "geneticist", also known as *father of genetics* was born in 1822 in Silisian, a village in Heizendorf (Austria). In 1843, he joined Augustinian monastry at Brunn (then in Austria, now Brno Czechoslovakia). In 1856, Mendel got interested in breeding of *Garden pea* (*Pisum sativum*). He selected pure breeding varieties or *pure lines of pea.* Breeding experiments were performed between 1859 – 1864. The results were read out in two meetings of *Natural History Society* of *Brunn* in 1865 and published in 1866 in "Proceedings of Brunn Natural History Society" under the topic "*Experiments in Plant Hybridisation*". Mendel died in 1884 without getting any recognition during his lifetime.

In 1900, *Hugo de Vries* of Holland, *Carl Correns* of Germany and *Erich von Tshermak* of Austria came to the same findings as were got by Mendel. Hugo de Vries found the paper of Mendel and got it reprinted in *'Flora'* in 1901. *Correns* converted two of the generalisations of Mendel into two laws of heredity. These are law of segregation and law of independent assortment.

(1) **Reasons for Mendel's success :** The reasons of his success can be discussed as follows:

(i) **Method of working :** He maintained the statistical records of all the experiments and analysed them. He selected genetically pure (pure breed line) and purity was tested by self-crossing the progeny for several generations.

(ii) **Selection of material :** Mendel selected garden pea as his experimental material because it has the following advantages.

It was an *annual plant.* Its short life–cycle made it possible to study several generations within a short period and has perfect *bisexual flowers* containing both male and female parts. The flowers are predominantly *self-pollinating* because of self-fertilization, plants are homozygous. It is, therefore, easy to get pure lines for several generations and also easy to cross because pollens from one plant can be introduced to the stigma of another plant by removing anthers (emasculation) and bagging. In addition to that there was one reason more for his success. He studied seven pairs of characters which were present on four different pairs of chromosomes.

(iii) **Selection of traits :** Mendel selected seven pairs of contrasting characters as listed in the table. Luckily all were related as dominant and recessive.

List of seven pairs of contrasting characters in pea plant

S.No.	Character	Dominant	Recessive
(1)	Stem length	Tall	Dwarf

(2)	Flower position	Axial	Terminal
(3)	Pod shape	Inflated	Constricted
(4)	Pod colour	Green	Yellow
(5)	Seed shape	Round	Wrinkled
(6)	Seed colour	Yellow	Green
(7)	Seed coat colour	Grey	White

(2) **Mendel's experiments**

(i) **Monohybrid cross :** Experiments with garden pea for *single pair* of contrasting characters.

(a) **Procedure :** Mendel crossed pure tall and dwarf plants. The plants belonged to $\mathbf{F_1}$ *generation* all tall were self-pollinated. The plants of $\mathbf{F_2}$ *generation* were both tall and dwarf, in approximate 3:1 ratio phenotypically and 1:2:1 genotypically. On, self-pollination, the tall plants of $\mathbf{F_2}$ only 1/3–rd breed true for tallness, the rest 2/3–rd produced tall and dwarf in the ratio of 3:1 **($\mathbf{F_3}$ *generation*)**. It means $\mathbf{F_2}$ generation consisted of three types of plants (instead of apparent two types) –

Tall homozygous (Pure)

1 25% **TT**

Tall heterozygous (Hybrid)

2 50% **Tt**

Dwarf homozygous (Pure)

1 25% **tt**

Hence it is to be said that in $\mathbf{F_2}$ generation 50% plants passes parental combination while 50% are new combination.

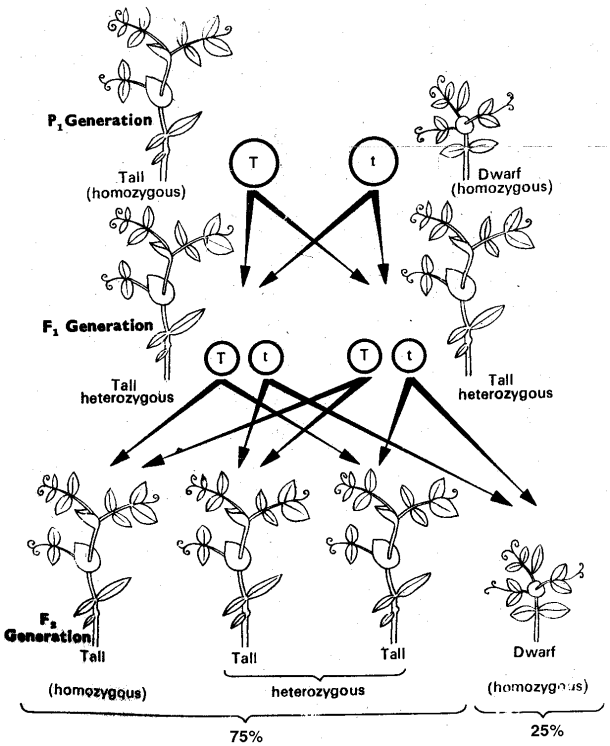


Fig : Mendel's monohybrids crosses between tall and dwarf pea plants

(b) **Mendel's explanation :** Mendel explained above results by presuming that *Tallness* and *dwarfness* are determined by a pair of contrasting factors or determiners (now these are called genes). A plant is tall because it possesses determiners for tallness (represented by **T**) and a plant is dwarf because it has determiners for dwarfness (represented by **t**). These determiners occur in pairs and are received one from either parent. On the basis of this behaviour the tallness is described as dominant character and dwarfness as recessive (*law of dominance*). The determiners are never contaminated. When gametes are formed, these unit factors segregate so that each gamete gets only one of the two alternative factors. When F_1 hybrids (**Tt**) are self pollinated the two entities separate out and unite independently producing tall and dwarf plants (*law of segregation*).

(ii) **Dihybrid cross** (Crosses involving *two pairs* of contrasting characters)

(a) **Procedure :** Later on Mendel conducted experiments to study the segregation and transmission of two pairs of contrasting characters at a time. Mendel found that a cross between round yellow and wrinkled green seeds **(P_1)** produced only round and yellow seeds in **F_1** generation, but in **F_2** four types of combinations were observed. These are

Round yellow 9 Parental combinations

Round green 3 Non-parental combinations

Wrinkled yellow 3 Non-parental combination

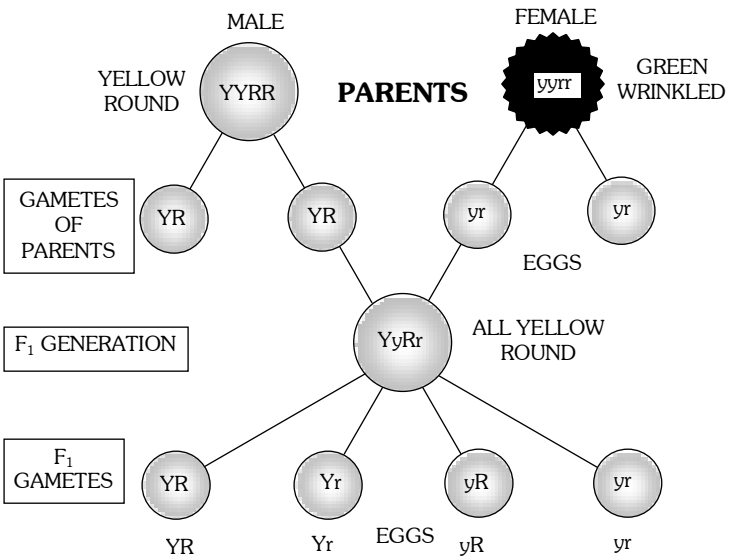
Wrinkled green 1 Parental combination

Thus the offsprings of F_2 generation were produced in the ratio of 9 : 3 : 3 : 1 phenotypically and 1 : 2 : 2 : 4 : 1 : 2 : 1 : 2 : 1 genotypically. This ratio is called *dihybrid ratio*.

The results can be represented as follows:

Mendel represented round character of seed by **R** and wrinkled by **r**. Similarly he designated the yellow character by **Y** and green by **y**. Therefore, it was a cross between **RRYY** and **rryy.**

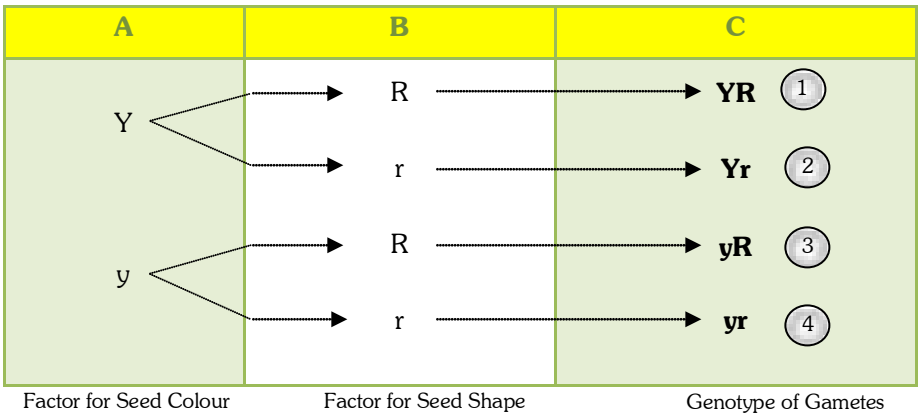
(b) **Mendel's explanation :** Mendel explained the results by assuming that the round and yellow characters are dominant over wrinkled and green so that all the **F_1** offsprings are round yellow. In **F_2**-generation since all the four characters were assorted out independent of the others, he said that a pair of alternating or contrasting characters behave independently of the other pair *i.e.*, seed colour is independent of seed coat.



POLLEN \ YR	YR	Yr	yR	yr
YR	YYRR YELLOW ROUND	YYRr YELLOW ROUND	YYRR YELLOW ROUND	YyRr YELLOW ROUND
Yr	YYRr YELLOW ROUND	YYrr YELLOW WRINKLED	YyRr YELLOW ROUND	Yyrr YELLOW WRINKLED
yR	YyRR YELLOW ROUND	YyRr YELLOW ROUND	yyRR GREEN ROUND	yyRr GREEN ROUND
yr	YyRr **YELLOW ROUND**	Yyrr YELLOW WRINKLED	yyRr GREEN ROUND	yyrr GREEN WRINKLED

Fig : Mendel's dihybrid-oross between pea plants having yellow round seeds and green wrinkled seeds

Therefore, at the time of gamete formation genes for round or wrinkled character of seed coat assorted out independently of the yellow or green colour of the seed. As a result four types of gametes with two old and two new combinations *i.e.*, **RY, ry Ry, rY** are formed from the **F_1** hybrid. These four types of gametes on random mating produced four types of offsprings in the ratio of 9:3:3:1 in **F_2** generation (*Law of Independent Assortment*).



(Forked-line method showing formation of four types of gametes from a F_1 – dihybrid for seed colour and seed shape)

(iii) **Trihybrid cross :** The offsprings shows 27 : 9 : 9 : 9 : 3 : 3 : 3 : 1 ratio is found in trihybrid cross. This suggests that a di, tri, or polyhybrid cross is actually a combination of respectively two, three or more monohybrid crosses operating together.

(iv) **Mendel's laws of inheritance :** Mendel's law are still true because these take place in sexually reproducing organisms or parents are of pure breeding. He enunciated two major laws of inheritance *i.e.*, law of segregation and law of independent assortment.

(a) **Law of segregation (Purity of gametes) :** The law of segregation states that when a pair of contrasting factors or genes or allelomorphs are brought together in a heterozygote (hybrid) the two members of the allelic pair remain together without being contaminated and when gametes are formed from the hybrid, the two separate out from each other and only one enters each gamete as seen in monohybrid and dihybrid cross. That is why the law of segregation is also described as law of purity of gametes.

(b) **Law of independent assortment :** If the inheritance of more than one pair of characters (two pairs or more) is studied simultaneously, the factors or genes for each pair of characters assort out independently of the other pairs. Mendel formulated this law from the results of a dihybrid cross.

Important Tips

- *Cytogenetics is Integrated study of cytology and genetics to find cytological basis for various events of genetics. This term was coined by Muller.*
- *H.J. Hammerlings proved that nucleus controls the heredity by a experiment on acetabularia (A unicellular green algae)*
- *Only sexually derived organisms are called **offspring** or **siblings** (offsprings at different births) e.g., brother and sister.*
- *Variations due to environment are known as **ecophenotypes**.*
- *Every test cross is **back cross** but every back cross is not a **test cross**.*
- *Back cross is used by breeders as a rapid method of making **homozygous**.*
- *When the two genetic loci produce identical phenotypes in cis and trans position they are considered to be **pseudoalleles**.*
- ***Somaclonal variations** are produced in tissue culture during differentiation of callus.*
- *In thalassemia, the β chain of haemoglobin is changed due to **frame shift mutation** as a result, bone marrow is not formed.*
- ***Bateson** coined the term **Genetics, allele, F_1, F_2, homozygous heterozygous** and **epistasis**. He is also known as **father of animal genetics**.*
- *The basis of genetic counseling is mendelism.*