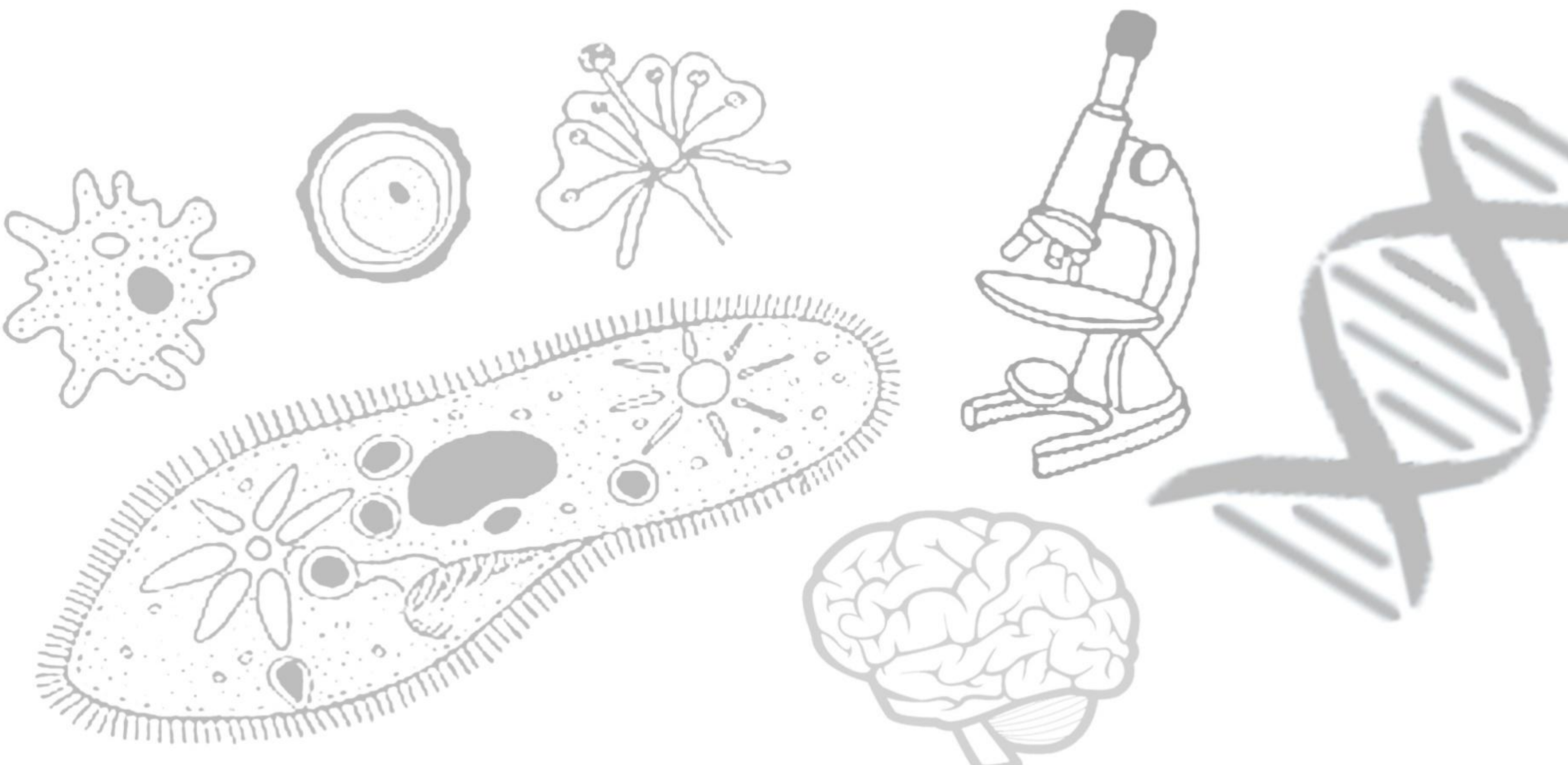
BIOLOGY



Mendel's Laws of Inheritance

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Heredity and Inheritance

- Heredity is the transmission of genetically based characters from parents to their offspring.
- Inheritance is the process by which characters or traits are passed on from one generation to another.
- Variation is the degree of differences in the progeny and between the progeny and the parents.

Why do variations occur?

In case of human beings, sexes are separate. Human beings reproduce through sexual reproduction. A haploid male gamete fuses with a haploid female gamete to form a diploid zygote with two sets of chromosomes. The zygote formed after fertilisation receives one set of chromosomes from the mother and the other set from the father. Thus, there is a recombination of genetic material, which ultimately results in variation in human beings.

Basic Features of Inheritance

Traits have alternative forms.

Traits are represented by discrete particulate entities which do not get blended or modified.

One alternative form of a trait can be expressed more often than the other.

An alternative form of a trait may remain hidden for one or more generations and then reappear in the unchanged state.

Particular forms of two or more traits may occur together in one generation and separate in subsequent generations.

Of the two alternative traits present together in an individual, only one gets expressed.

DID YOU KNOW ?

The branch of biology which deals with the study of heredity and variation is called genetics.

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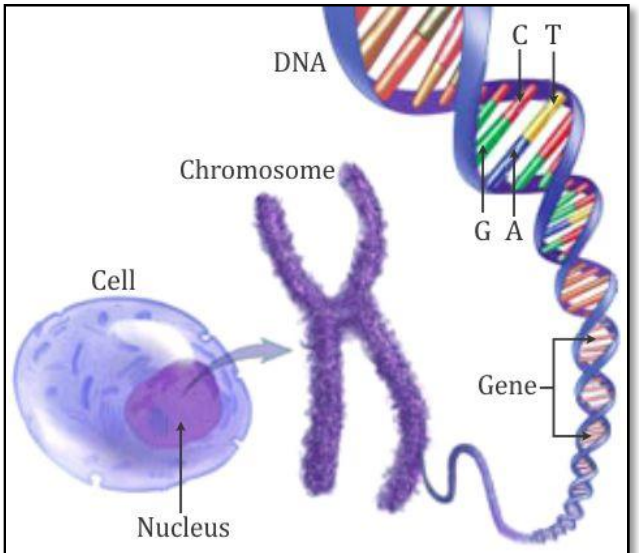
- Genes are specific parts of chromosomes or deoxyribonucleic acid (DNA) segments which determine the hereditary characteristics.
- Genes are segments of DNA called cistrons.



Every chromosome has small parts called genes

- Normally, every gene has two alternative forms for a character, each of which produces different effects in an organism. These alternative forms are called alleles. Example: In pea plants, the stem height is controlled by two alleles, one for tallness and the other for dwarfness.
- Of the two alleles of a gene, one is dominant, i.e. super-ruling and the other is recessive, i.e. subordinate or submissive. A dominant allele is an allele which hides or masks the expression of its corresponding allele which then becomes recessive.
- The dominant and recessive alleles are represented differently. According to the rules of genetics, the dominant allele is represented by a capital letter and the recessive allele is represented by a small letter of the alphabet. In pea plants, the dominant allele for tallness is T and the recessive allele for dwarfness is t.
- The genetic constitution of an organism is called genotype. It is the description of genes present in an organism. The genotype is always denoted by a pair of letters. Thus, the genotype of a tall plant could be TT or Tt, while that of a dwarf plant is tt.
- Phenotype refers to the observable characteristic or the expressed shown character of an organism. Example: Being 'tall' or 'dwarf' are the phenotypes of a plant because these traits are visible to us. The phenotype of an organism is determined by its genotype. Example: Genotype TT or Tt results in a tall phenotype and genotype tt results in a dwarf phenotype.
- When two parents are crossed to produce progeny, their progeny is called first filial generation or F1 generation. When the first generation progeny or F1 progeny is crossed among themselves to produce a second generation, then this progeny is called second filial generation or F2 generation.
- A new form of a plant resulting from a cross of different varieties of a plant is known as a hybrid.

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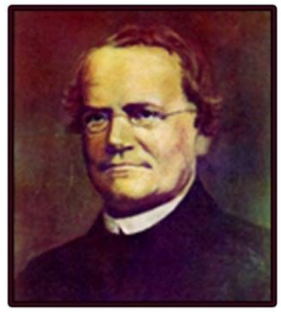
DID YOU KNOW ?

W. Johannsen coined the term pure line (1900), gene (1909), genotype and phenotype (1911).

About the scientist



Sir Gregor Johann Mendel

Sir Gregor Johann Mendel (1822-1884) was not only a priest in a monastery, but also a naturalist. He conducted several experiments in the monastery garden to study transmission of certain traits in pea plants. Mendel was the first scientist to make a systematic study of patterns of inheritance, which involved the transfer of characteristics from parents to the progeny. He is now recognized as the “Father of Genetics” due to his remarkable contribution to the field. Several other scientists had studied the inheritance of traits. However, Mendel blended his knowledge of science and mathematics and was the first person to give the hypothesis of the existence of a factor called the Mendelian factor to explain continuity and variation. This factor is now known as “gene”. The significance of Mendel’s work was not recognized until the turn of the 20th century. In 1900, scientists rediscovered Mendel’s work and its importance in the field of genetics.

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Reasons for Mendel's Success

- Selected only pure breeding varieties of pea for his experiments.
- Took only those traits which did not show linkage, interaction or incomplete dominance.
- Characters chosen had distinctive contrasting traits.
- Took only one or two characters at a time for study.
- Studied the inheritance of a character for 3 or more generations.
- Performed reciprocal crosses and raised a large number of progenies.
- Took care to avoid contamination with unwanted pollen.
- Kept a complete record of every cross, subsequent self-breeding and number of seeds produced.
- Formulated theoretical explanations for interpretation of results.
- Used statistical methods and law of probability for analysis of results.
- Luckily selected only those traits, the genes of which did not interact.
- Did not attempt to explain all the variations found in his results.

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Mendel's Experiments

- Mendel conducted experiments on the pea plant (*Pisum sativum*) and studied the inheritance of certain traits.

Traits	Shape of seeds	Colour of seeds	Colour of pods	Shape of pods	Plant height	Position of flowers	Flower colour
Dominant trait	Round	Yellow	Green	Full	Tall	At leaf junction	Purple
Recessive trait	Wrinkled	Green	Yellow	Flat, constricted	Short	At tips of branches	White

Seven pairs of contrasting traits in pea plant

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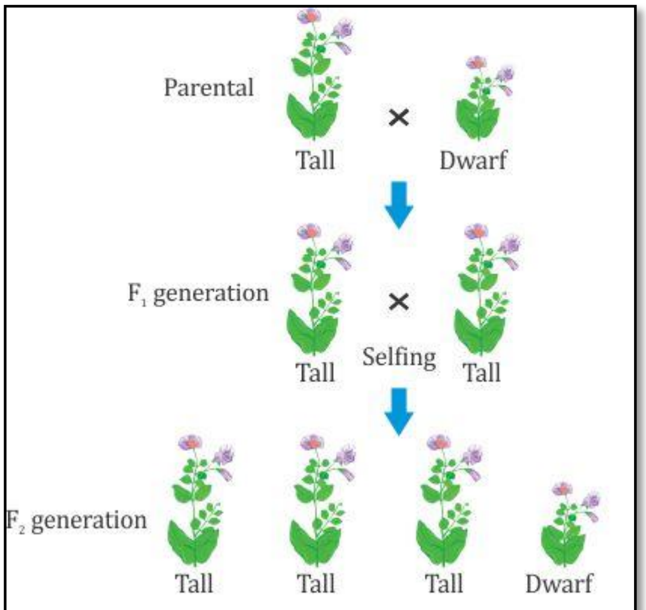
One Gene Inheritance (Monohybrid Cross)

- A cross which involves only a single pair of contrasting characters is called a monohybrid cross.
- Consider pea plants with a pair of contrasting characters—tallness and dwarfness with respect to the height of the stem. As pea plants are self-fertilising, Mendel bred them for several generations till he obtained pure varieties. The parental generation P_1 has the genotype TT for a pure tall parent plant and tt for a pure dwarf plant. The two parents are homozygous, having one type of gamete each, i.e. T from tall parent and t from dwarf parent. These two plants are crossed with one another.
- Mendel found that all plants were tall. He called them first filial or F_1 generation seeds. The F_1 generation has genetic constitution Tt. It is genotypically a hybrid and a heterozygous plant with two different alleles. Phenotypically, the plant is tall because the allele or the gene T for tallness masks the effect of its corresponding recessive gene t.



An illustration of monohybrid cross

- F_1 plants are self-pollinated to obtain the F_2 generation, $F_1 \times F_1 = F_2$. Now, each of these parents is heterozygous tall with the genotype Tt. So, we have Tt × Tt. The gametes produced from each of the parents are of two types, T and t.
- The second filial generation F_2 has a genotypic ratio of 1 TT : 2 Tt : 1 tt. In this case, as the allele T for tallness is dominant, the pea plants with the genotype Tt will be tall. The phenotypic ratio is 3 tall : 1 dwarf. Genotypically, it shows three types of plants: 1 TT, which is homozygous tall, 2 Tt which are heterozygous tall and 1 tt which is homozygous dwarf. Thus, the genotypic ratio is 1 TT : 2 Tt : 1 tt.
- The ratio of 3 : 1 is known as the monohybrid ratio. The results of a monohybrid cross enabled Mendel to formulate his first law of inheritance which is called the law of segregation.

Law of Segregation

The characteristics or traits of an organism are determined by internal 'factors' which occur in pairs. Only one of a pair of such factors can be present in a single gamete.

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Differences between Homozygous and Heterozygous Individuals

Homozygous individual	Heterozygous individual
1. Pure for a trait and breeds true	1. Seldom pure and produces offspring with different genotypes on selfing
2. Both alleles for a character are similar	2. Carries dissimilar alleles
3. Can carry either dominant or recessive alleles but not both	3. Has both dominant and recessive alleles
4. Produces gametes of one type	4. Produces gametes of two types
5. Does not exhibit extra vigour	5. Can exhibit extra vigour called hybrid vigour or heterosis

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Two Gene Inheritance (Dihybrid Cross)

- A cross which involves plants with two pairs of contrasting characters is called a dihybrid cross.
- Consider a cross of pea plants with round and yellow seeds and plants with wrinkled and green seeds. In this cross, the two pairs of characters involved are seed shape and seed colour.
- The capital letter R is used to denote round-shaped seeds and the capital letter Y to denote yellow-coloured seeds. The genotype of the parent plant is RRYY. The other parent has wrinkled seeds which are green. The small letter r denotes wrinkled seeds and the small letter y denotes green-coloured seeds. The genotype of the parent is rryy. The two parent plants are then crossed, RRYY × rryy.
- When the gametes of the P_1 generation are formed, each pair of alleles segregates independently of the other. Hence, during gamete formation, RRYY plants produce gametes RY. The gametes RR or YY are not produced by them. Similarly, the other parent rryy produces gametes ry. The gametes rr or yy are not produced by them.
- In each of the gametes, a pair of alleles is represented by only one of its members. In the P_1 generation, the parent RRYY produces 2 gametes, RY and RY, and parent rryy produces 2 gametes, ry and ry.
- Mendel observed that the F_1 plants were with round yellow seeds (phenotypically). The genotype of these F_1 hybrids was RrYy. The genotype shows R which is dominant over r with respect to seed shape and results in round seeds. The genotype also contains Y which is dominant over y with respect to seed colour and results in yellow-coloured seeds. All the F_1 plants are called dihybrids as they have two sets of contrasting characters and have the genotype RrYy.
- The F_1 plants produced four types of gametes—ry, RY, rY and Ry—during gamete formation. On the other hand, the parental plants P_1 produced only two types of gametes RY and ry. The gametes produced by F_1 plants which are similar to those produced by P_1 plants are called parental combinations. The two other types of gametes rY and Ry produced by F_1 plants during gamete formation are called recombinations.
- When the F_1 plants are self-pollinated, they produce a new generation of plants known as F_2 or the second filial generation. The four types of male gametes and the four types of female gametes produce 16 different mating combinations in all. The 16 different types of F_2 individuals can be easily represented with the help of a Punnett square.
- The F_2 generation obtained by self-pollination of F_1 plants exhibits 16 plants with different genotypes. Of a total of 16 plants, 9 plants are with round and yellow seeds RRYY, 3 are with wrinkled and yellow seeds rrYY, 3 are with round and green seeds RRyy and 1 is with wrinkled and green seed rryy. Thus, the phenotypic ratio of F2 plants is 9 : 3 : 3 : 1.
- Genotypically, there are 9 different types of plant combinations, RRYY, RrYy, Rryy, rryy, RRYy, rrYy, Rryy, Rryy and rrYY. The genotypic ratio is 1 : 4 : 1 : 1 : 1: 2 : 2 : 2 : 2.
- The genotype RRYY is a pure dominant with homozygous round and homozygous yellow seeds. The genotype rryy is a pure recessive with homozygous wrinkled and homozygous green seeds.
- The 9:3:3:1 ratio of each phenotype of the seeds in the F_2 generation is called dihybrid ratio. The results of the dihybrid ratio enabled Mendel to formulate his second law of inheritance which is called the law of independent assortment.

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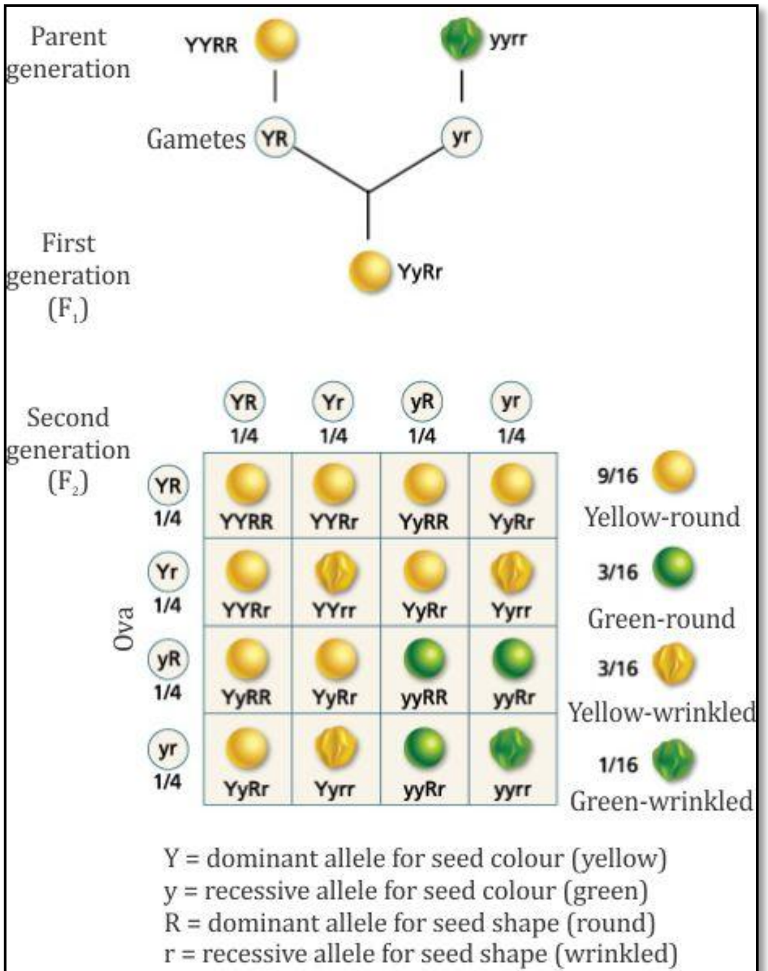

An illustration of dihybrid cross

Law of independent assortment

In the inheritance of more than one pair of traits in a cross simultaneously, the factors responsible for each pair of traits are distributed independently to the gametes.

Differences between Monohybrid and Dihybrid Crosses

Monohybrid Cross	Dihybrid Cross
1. It is a cross between two pure organisms in order to study the inheritance of a single pair of alleles.	1. It is a cross between two pure organisms in order to study the inheritance of two pairs of alleles.
2. It produces a phenotypic monohybrid ratio of 3:1 in the F_2 generation.	2. It produces a phenotypic dihybrid ratio of 9:3:3:1 in the F_2 generation.
3. It produces a genotypic ratio of 1:2:1 in the F_2 generation.	3. It produces a genotypic ratio of 1:2:1:2:4:2:1:2:1 in the F_2 generation.

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Post-Mendelian Discoveries

- Gene interaction is the influence of alleles and non-alleles on the normal phenotypic expression of genes.

- Gene interaction
  - Intragenic interaction
    - Incomplete dominance
    - Codominance
    - Multiple alleles
  - Intergenic interaction
    - Epistasis
    - Duplicate genes
    - Complementary genes
    - Supplementary genes
    - Lethal genes
    - Inhibitory genes

- In intragenic or interallelic interaction, the two alleles of a gene interact in such a way as to produce a phenotypic expression different from the typical dominant–recessive phenotype. Examples: Incomplete dominance, codominance, multiple alleles
- In intergenic or non-allelic interaction, two or more independent genes present on the same or different chromosomes interact to produce a different expression. Examples: Epistasis, duplicate genes, complementary genes, supplementary genes, lethal genes, inhibitory genes

Incomplete Dominance

- Incomplete dominance is the phenomenon where none of the two contrasting alleles or factors is dominant.
- The expression of the character in a hybrid or F_1 individual is intermediate or a fine mixture of the expression of the two factors.
- Incomplete dominance is an example of quantitative inheritance where only a single gene pair is involved.
- The phenotypic ratio of offspring in the F_2 generation is 1:2:1 which is similar to the genotypic ratio.

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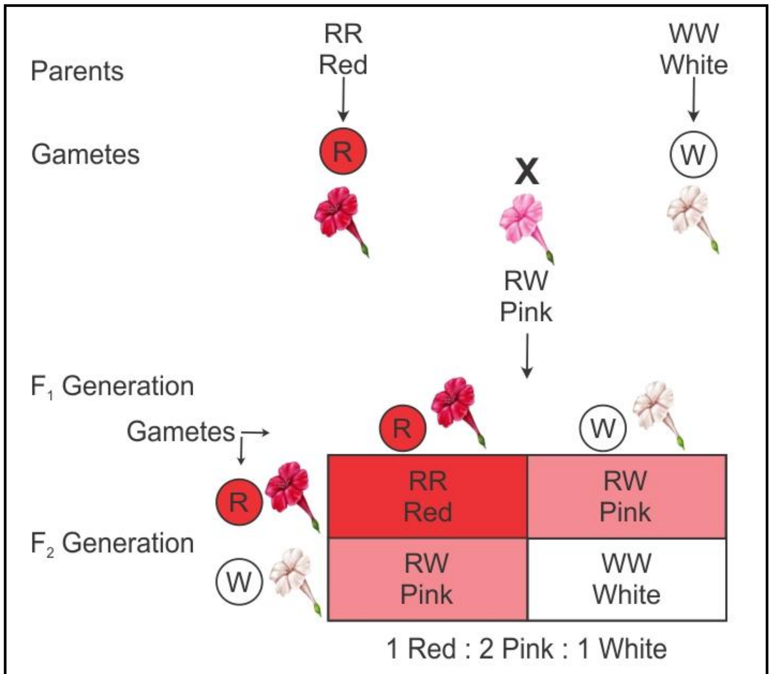
Example of Incomplete Dominance

- In *Mirabilis jalapa* (Four o'clock plant) and *Antirrhinum majus* (Snapdragon plant), there are two types of flowers, red and white.
- When the two types of plants are crossed, the hybrids of the F_1 generation bear pink flowers.
- When the F_1 hybrids are selfed, we obtain three types of plants in the F_2 generation—one with red flowers, one with pink flowers and one with white flowers in the ratio of 1:2:1.
- The appearance of pink colour is either due to the mixing of red and white colours or the expression of a single gene for a pigmented flower which produces only pink colour.



Incomplete dominance in *Mirabilis jalapa*

Differences between Dominance and Incomplete Dominance

Dominance

- F_1 hybrid is similar to the parent.
- Phenotypic ratio is different from genotypic ratio.
- In the F_1 hybrid, the dominant trait is completely expressed.
- A single dominant allele is expressed completely.

Incomplete Dominance

- F_1 hybrid is different from either of the two parents.
- Phenotypic and genotypic ratios are the same.
- In the F_1 hybrid, the dominant trait is incompletely expressed.
- Two alleles are required for the expression of a completely dominant trait.

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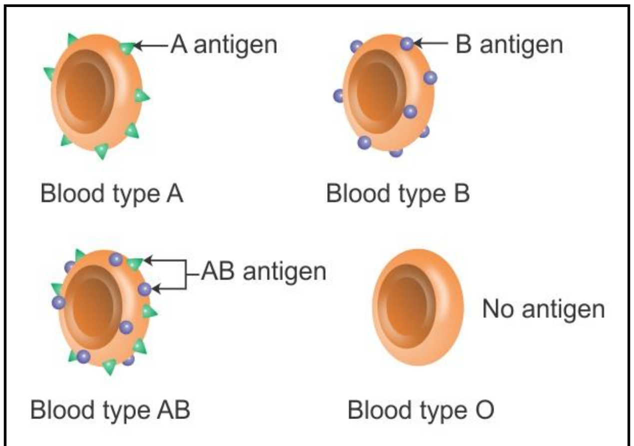
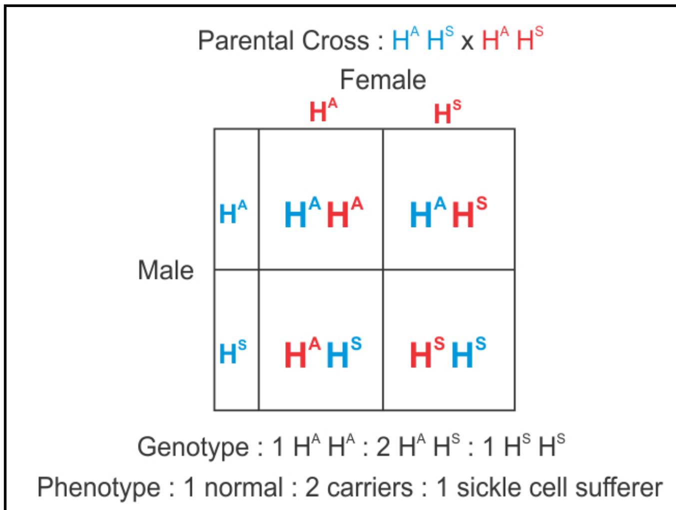
Codominance

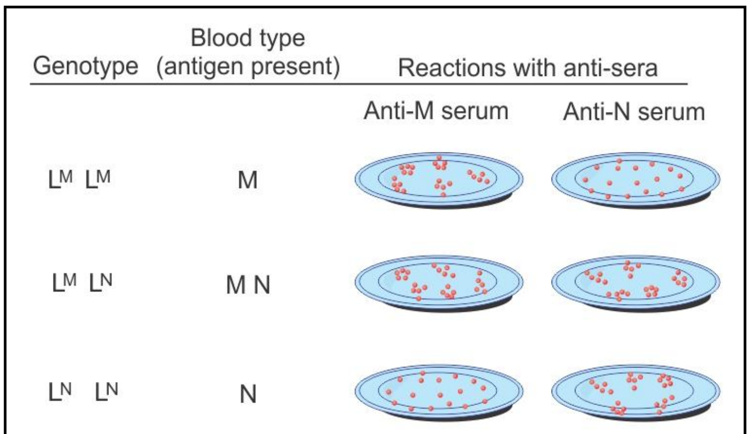
- The phenomenon of expression of both alleles in a heterozygote is called codominance.
- The alleles which do not show a dominant–recessive relationship and are able to express themselves independently when present together are called codominant alleles.
- The heterozygous condition has a phenotype different from either of the homozygous genotypes.
- The joint character may appear to be intermediate between the ones produced by the two homozygous genotypes.
- Examples: AB blood group, MN blood group, sickle cell haemoglobin



Codominance (AB blood group)



Codominance (Sickle cell anaemia)



Codominance (MN blood group)

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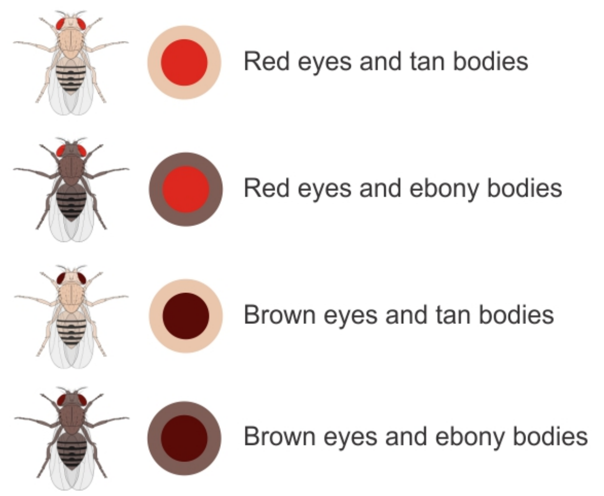
Multiple Alleles

- Multiple alleles refer to the presence of more than two alleles of a gene.
- They are produced due to repeated mutation of the same gene but in different directions.
- Examples: Eye colour in Drosophila, self-incompatibility in some plants



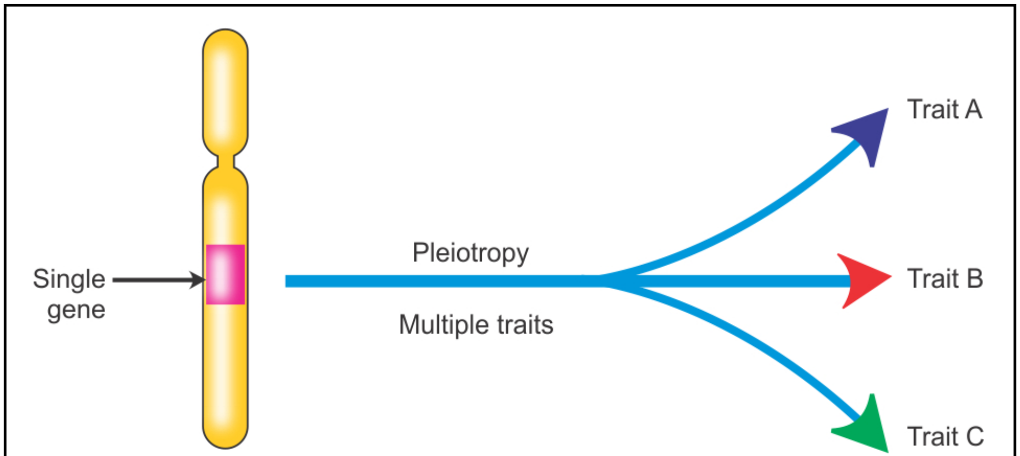
Multiple alleles (Eye colour in *Drosophila*)

DID YOU KNOW ?

Bernstein (1924, 1925) discovered multiple alleles, codominance and dominant-recessive relationships in determination of human blood groups.

Pleiotropy

- The ability of a gene to have multiple phenotypic effects as it influences several characters simultaneously is known as pleiotropy.
- The gene with a multiple phenotypic effect because of its ability to control expression of two or more characters is called pleiotropic gene.



Pleiotropy

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- Pleiotropy occurs because of the effect of the gene on two or more interrelated metabolic pathways which contribute to the formation of different phenotypes.
- Examples: In garden pea plant, the gene which controls the flower colour also controls the colour of the seed coat and the presence of red spots in the leaf axils.

DID YOU KNOW ?

Marfan's syndrome is caused in human beings by a pleiotropic gene which is characterised by slender body, limb elongation, hypermobility in joints, lens dislocation and tendency to develop heart diseases.

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