Variations: the differences in the characters (traits) among the individuals of a species are called variations. Variations may also be produced during asexual mode of reproduction (due to small inaccuracies in copying of DNA), but sexual reproduction produces a large number of variations, e.g. in plants, less variations are seen in comparison to animals which reproduce mainly by sexual reproduction.

Variations are beneficial and necessary for organic evolution. These variations may environmental or genetic.

Types of Variations: There are two types of variations—germinal and somatic.

1. Germinal, or genetic, variations: are caused either by differences in the numbers or structures of chromosomes or by differences in genes (units of heredity). Changes in genes are the primary sources of germinal variations. These variations are heritable. Height and eye colour are examples of germinal variations.
2. Somatic variations: result from several factors such as climate, food supply, environment, and interactions with other organisms. These variations are not due to changes in genes or chromosomes. They are not transmittable to future generations. Hence, they are not significant in the process of evolution.

Heritable Variation: Heredity involves the transmission of characteristics from parents to their offspring. Among sexually reproducing organisms, the progeny are not exact duplicates of their parents.

Heritable variation is the result of changes in the arrangement of genes on the chromosomes and changes in the sequence of the bases that constitute DNA. Such changes occur spontaneously and randomly in the DNA, which undergoes duplication just before the cell divides.

Duplication is done with great precision, but once in about ten million duplications there is an error as a different base may be put in place of the correct one. This mistake, or mutation, gets copied in subsequent cell divisions. Such spontaneous changes in the DNA are proportional to the number of duplications and cell divisions. With successive generations these variations go on accumulating in the descendants. Hence, those organisms that reproduce at a faster rate accumulate a greater number of variations.

Accumulation of Variations: Variations in an individual may be an advantage or a disadvantage for it. An advantage of sexual reproduction is that the variations accumulated in the gametes of each sex are combined when they fuse to form the zygote. Hence an offspring produced from the zygote receives and carries the variations of both the parents. On the other hand in asexual reproduction there are minor differences among the offspring. These are due to small errors in DNA copying. As gametes and zygote formation are not involved, the asexually produced offspring are quite similar. They have fewer variations.

Significance of Variation:

1. They form the basis of heredity
2. Adaptability to adverse conditions is brought about by variations
3. New varieties of organisms may arise due to the genetic variation laying the foundation for evolution.

Heredity:

1. The traits, or characteristics, of a species are passed down from one generation to another.

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2. The closer the relationship, the greater is the similarity. This continuity of traits, which is maintained for all species and passed down to succeeding generations, is called heredity. However, along with the similarities between brothers and sisters, there is also some dissimilarity. The dissimilarities are called variations. The study of the pattern of transmission of characters from parents to their offspring is called genetics.

Both father and mother pass on genes, inherited from their own parents, to their children. Genes are stretches of DNA containing coded information for making proteins. These are found on the chromosomes of cells. Genes are the units of heredity. The genetic constitution of an individual organism is called its genotype. The genotype determines the characteristic features of an individual (such as height, complexion, hair colour, etc.), which are slowly expressed during development. These characteristic features of an individual also depend on the environment. The sum of the activities of genes and the impact of the environment on them leads to the formation of visible traits, called phenotype.

Mendel's Laws for the Inheritance of Traits: Gregor Johann Mendel (1822—84) was an Austrian monk and botanist. He is regarded as the father of genetics as some of the basic laws of inheritance were proposed by him. In 1857, Mendel began a series of experiments on the pea plant (Pisum sativum) to study the pattern of inheritance of various characters. He chose pea plants for three reasons.

1. Pea plants are self-pollinating.
2. They are easy to cultivate.
3. They have sharply defined characters.

Mendel chose to study seven different characters in pea plants. Each of these characters such as height, seed shape, seed colour, etc., had two sharply defined and contrasting traits (e.g., tallness and dwarfness, round seed and wrinkled seed, yellow seed and green seed). He crossed a variety of pea plant carrying a particular trait (e.g., tallness) of a character (such as height) with another variety having a contrasting trait (e.g., dwarfness) of the same character.

These two plants were considered as the parental generation (P). The generation that was produced by crossing these two was called the first filial generation (F_1). When F_1 plants were crossed among themselves, the generation that was produced was called the second filial generation (F_2). The results of Mendel's experiments showed the following:

1. Whenever two traits of a character were crossed, the F_1 plants showed only one of the traits; the other trait never appeared. It did not matter whether the trait came from the pollen or the egg.
2. The trait that did not appear in F_1 reappeared in F_2, but in 1/4 of the total number of plants.

He explained that each genetic character was represented controlled by a pair of unit factors. These unit factors came to be known as alleles. When the term 'gene' was coined and defined, the allele became synonymous with the gene. One of the alleles came from one parent and the other from the other parent. The first-generation plants of Mendel's experiment were all tall plants. But the allele representing dwarfness was neither destroyed nor altered. It could not be expressed in the first generation because it was dominated by the allele

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representing tallness. In other Words, the allele for tallness was dominant and the allele for dwarfness was recessive.

Notations used in Mendel's experiments: The dominant trait is usually represented by a capital letter. For example, tallness is represented by 'T' and dwarfness is represented by the corresponding small letter 't'. If tallness is due to a pair of dominant alleles, it is written as TT. If tallness is due to only one dominant allele, it is written as Tt. If both the alleles are recessive, making the organism dwarf, then it is written as tt. A homozygous condition is one in which both the alleles are of the same nature, e.g., TT or tt. A alleles are of different nature, e.g., Tt. When two heterozygous condition is one in which the characters are taken into account, the notation for the homozygous dominant could be AABB, and for the homozygous recessive it could be aabb.

Term	Definition
Gene	A functional unit of heredity present on chromosomes of cell nucleus. It is composed of DNA and codes for one polynucleotide (protein). It determines a particular character (phenotype).
Allele	One of the different forms of a particular gene, e.g. hair colour.
Dominant allele	An allele, whose phenotype will be expressed even in the presence of another allele of that gene. It is represented by a capital letter, e.g. T.
Recessive allele	An allele, which gets masked in the presence of dominant allele and can only affect the phenotype in the absence of a dominant gene. It is represented by a small letter, e.g. t.
Genotype	Genetic composition of an individual.
Phenotype	The expression of the genotype, which is an observable or measurable characteristic.
Punnett square	Probability diagram, illustrating the possible offsprings of mating.
Chromosome	A long rod-like structure in a nucleus. It appears during cell division and is thought to carry genes.
Hybrid	An individual having two different alleles for the same trait.
Monohybrid cross	A hybridisation cross in which inheritance of only one pair of contrasting characters is studied.
Dihybrid cross	A cross in which inheritance of two pairs of contrasting characters is simultaneously studied.
Homozygous	A condition in which an individual possesses a pair of identical genes controlling a given character and will breed true for this character (e.g. occurrence of two identical alleles for tallness in a P_1 tall pea plant).
Heterozygous	A condition in which an individual has a pair of contrasting genes for any one character and will not breed true for this character (e.g. existence of dominant and recessive alleles in F_1 hybrid tall pea plant).
Progeny	A descendant or offspring as a daughter organism.
Gametes	Reproductive cells containing only one set (haploid) of dissimilar chromosome.

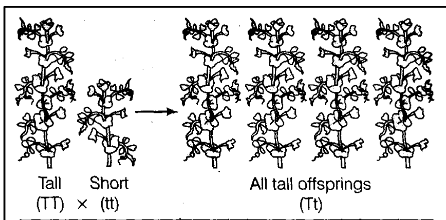
He performed following two experiments:

1. Inheritance of Traits for one Contrasting Character:

Mendel took pea plants with different characteristics such as height (tall and short plants).

• The progeny produced from them (F1 -generation plants) were all tall. Mendel then allowed F1 progeny plants to undergo self-pollination.

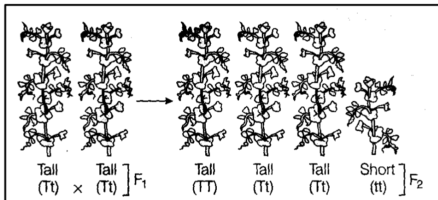
• In the F2-generation, he found that all plants were no three quarter were tall and one quarter of them were short. This observation indicated that both the traits of shortness and tallness were inherited in F1 -generation, but only the tallness trait was expressed in F1 -generation. This is known as the law of dominance.



Mendel's experiment showing law of dominance

• Two copies of the traits are inherited in each sexually reproducing organism.

TT and Tt are phenotypically tall plants, whereas tt is a short plant. For a plant to be tall, the single copy of 'T' is enough. Therefore, in traits Tt, 'T' is a dominant trait while 't' is a recessive trait. In F2 - generation, both the characters are recovered, though one of these is not seen in F1 stage. During gamete formation, the factor or allele of a pair segregate from each other. This is known as the segregation.

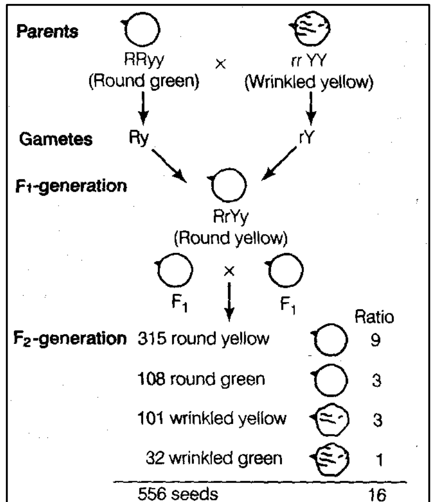


Mendel's experiment showing law of segregation

<u>2.Inheritance of Traits for Two Visible Contrasting Characters:</u> Mendel took pea plants with two contrasting characters, i.e. one with a green round seed and the other one with a yellow wrinkled seed.

- When the F1 progeny was obtained they had round and yellow seeds, thus establishing that round and yellow are dominant traits.
- Mendel then allowed the F1 progeny to be self-crossed (self-pollination) to obtain F2 progeny. He found that seeds were round yellow, round green, wrinkled yellow and some were wrinkled green.
- The ratio of plants with above characteristics was 9:3:3:1, respectively (Mendel observed that two new combinations had appeared in F2).

In F2-generation, all the four characters were assorted out independent of the others. Therefore, he said that a pair of alternating or contrasting characters behaves independently of the other pair, i.e. seed colour is independent of seed coat. This is known as the law of independent assortment.



Independent inheritance of two separate traits, shape and colour of seeds

Law	States That	Parent Cross	Offspring
Dominance	When parent plants are pure for contrasting traits, only one form of the trait will appear in the next generation, that is known as dominant trait (T). The hidden trait (t) is recessive.	TT × tt (Tall) (Short)	Tt (All tall)
Segregation	In F_1 hybrid, the dominant and recessive traits though remain together for long time but do not mix with each other and separate or segregate at the time of gamete formation.	Tt × Tt (Tall) (Tall)	3 : 1 (Tall Short)
Independent Assortment	The inheritance of one character is always independent of the inheritance of other character within the same individual.	RRyy × rrYY (Round green, wrinkled yellow)	9 : 3 : 3 : 1 (Round yellow : round green : wrinkled yellow : wrinkled green)

<u>Mechanism of expression of traits</u>: A gene contains the information for making proteins in the cell. The proteins synthesized according to this information may be enzymes that catalyse biochemical reactions. Each trait is the outcome of several such biochemical reactions, each of which is controlled by a specific enzyme. If the enzyme is not produced because its gene is absent, that particular reaction will not occur and the trait resulting from its reaction will not appear phenotypically. Thus, each trait is controlled by a gene. Each parent contributes one copy of the gene for a particular character. Thus there are two genes for every character. In the gamete, however, only one copy is present because of reduction division. What Mendel perceived was that each gene (allele) is an

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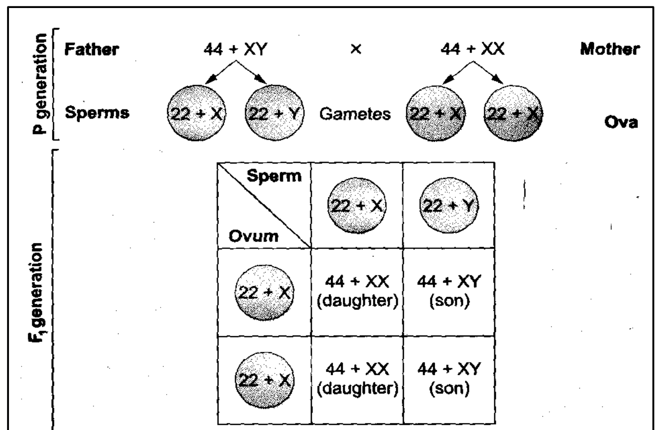
independent unit which is neither linked with nor influenced by the other gene. Also, each allele can be separated in gametes.

Mechanism of Inheritance: If both parents help to determine the trait in the progeny, both parents must be contributing a copy of the same gene.

- Thus, each pea plant must have two sets of all genes, one inherited from each parent. So, each germ cell must have only one gene set.
- Each set of gene is present not as a single thread of DNA, but as separate independent pieces called chromosome and each cell will have two copies of each chromosome, one inherited from each parent, i.e. one from male parent and one from female parent.
- When two germ cells combine, they will restore the normal number of chromosomes in the progeny, ensuring the stability of the DNA of species. Such mechanism of inheritance explains the result of Mendel's experiments and is used by all sexually and asexually reproducing organisms.

Sex Determination: A person can have either a male sex or a female sex. The process by which sex of a newborn individual is determined is called sex determination. There are different strategies by which sex is determined in different species. In some species, environmental factors are important in determining the sex of the developing individual.



In human beings, sex is determined by genetic inheritance. Genes inherited from the parents determine whether an offspring will be a boy or a girl. Genes for all the characters are linearly arranged on chromosomes. These include the genes for sexual characters. Generally, characters related to the reproductive system are called sexual characters and those that are not are called vegetative characters. The chromosomes that carry genes for sexual characters are called sex chromosomes, while those that carry genes for the vegetative characters are called autosomes.

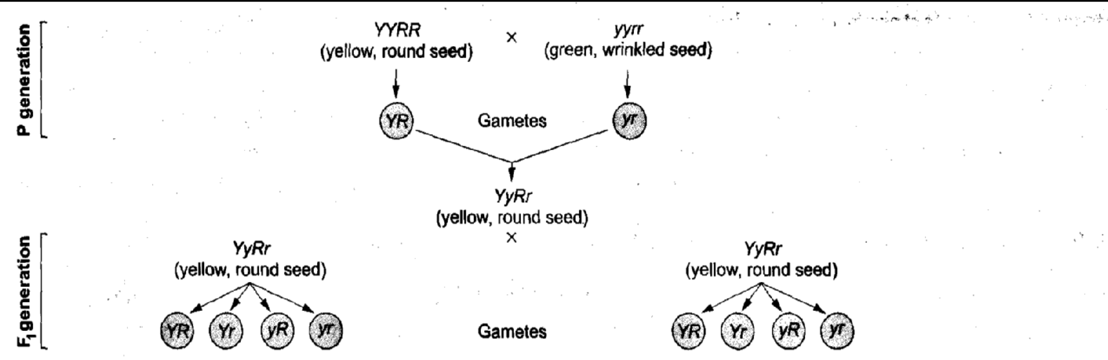
A sex chromosome that carries the genes for male characters is called Y chromosome and one which carries the genes for female characters is called X chromosome. We have a total of 46 chromosomes. Half of them come from the mother and the rest, from the father.

Out of these 46 chromosomes, 44 are autosomes and 2 are sex chromosomes. The sex chromosomes are not always a perfect pair. In females there are 44 autosomes and two X chromosomes. In males there are 44 autosomes, one X chromosome and one Y chromosome. So the chromosomes in woman are 44+ XX, while the chromosomes in man are 44+ XY.

All children obtain either chromosome from both parents. Females have a perfect pair of sex chromosome (homogametic) and thus, contribute X-chromosome to both the sexes of progeny but males have a mismatched pair (heterogametic) in which one is X (normal sized) and the other is V-chromosome (short in size).

Hence, an egg fertilised by X carrying sperm results in a zygote with XX, which becomes a female and if an egg is fertilised by Y carrying sperm, it results in a XY zygote that becomes male. Thus, the sex of the children will be determined by what they inherit from their father. A child who inherits an X-chromosome will be a girl and one who inherits a V-chromosome will be a boy.

<u>Role of environment in sex determination</u>: Environmental conditions such as temperature around the developing embryo may determine sex in some animals. Such conditions may override the genetic basis, Some animals such as snails can even change their sex, showing that their sex is not genetically determined. Incubation of the eggs of the turtle *Chrysema picta* at a high temperature produces females. But the incubation of the eggs of the lizard *Agama agama* at a high temperature produces males.



F_2 generation

Gametes	YR	Yr	yR	yr
YR	YYRR (yellow, round seed)	YYRr (yellow, round seed)	YyRR (yellow, round seed)	YyRr (yellow, round seed)
Yr	YYRr (yellow, round seed)	YYrr (yellow, wrinkled seed)	YyRr (yellow, round seed)	Yyrr (yellow, wrinkled seed)
yR	YyRR (yellow, round seed)	YyRr (yellow, round seed)	yyRR (green, round seed)	yyRr (green, round seed)
yr	YyRr (yellow, round seed)	Yyrr (yellow, wrinkled seed)	yyRr (green, round seed)	yyrr (green, wrinkled seed)

Genotype ratio
YYRR : YYRr : YyRR : YyRr : YYrr : Yyrr : yyRR : yyRr : yyrr
= 1 : 2 : 2 : 4 : 1 : 2 : 1 : 2 : 1

Phenotype ratio
(yellow, round seeds) : (Yellow, wrinkled seed) : (green, round seeds) : (green, wrinkled seeds)
= 9 : 3 : 3 : 1

How Blood Groups are Inherited

A person has one of the four blood groups : A, B, AB or 0. This blood group system is controlled by a gene which has three different forms denoted by the symbols I^A, I^B and I^O. The genes I^A and I^B show no dominance over each other, that is, they are codominant. However genes $I^A$and I^B both are dominant over the gene I^O. In other words, the blood gene I^O is recessive in relation to genes I^A and I^B.

Although there are three genes (or three gene forms) for blood, but any one person can have only two of them. So, the blood group of a person depends on which two forms of the genes he poses.

If the genotype (gene combination) is $I^A I^A$, then the blood group of the person is A. And if the genotype is $I^A I^O$ even then the blood group is A (because I^O is a recessive gene).

(ii) If the genotype is $I^B I^B$, then the blood group of the person is B. And if the genotype is $1^B I^O$ even then the blood group is B (because I^0 is a recessive gene)

(iii) If the genotype is $I^A I^B$, then the blood group of the person is AB.

(iv) If the genotype is $I^O I^O$, then the blood group of the person is 0.

Let us solve one problem now.

Sample Problem. A man having blood group A marries a woman having blood group O they have a child. What will be the blood group of the child?

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Answer. The answer to this question depends on whether the blood group A of the m gene combination $I^A I^A$ or $I^A I^O$

(i) **When the blood group A** has **genotype $1^A 1^A$** In this case the genotype of man's blood is $I^A I^A$ and that of woman's blood is **$I^0 I^0$.** So, the child will have blood group A (because the gene I^A is dominant over gene I^O).

(ii) **When the** blood **group A** has genotype I^A **I^0** Here the genotype of man's blood is $I^A I^O$ and that of woman's blood is **$I^0 I^0$.** So, in this case there is an equal chance that the genotvpe child's blood can be either $I^A I^0$ or **$I^0 I^0$.** Due to this, there is an equal chance of the child acquiring blood group A or blood group 0.

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