

Genetics - Some basic fundamentals ^①

Genetics: It is the study of transmission of characters from parents to offspring and the laws relating to such transmission.

Heredity: The phenomenon of passing of characters from parents to progeny through successive generations is called heredity.

Variation: The variation in traits or characteristics among individuals within a species is termed variations. For eg:
In the majority of individuals, the earlobe is free-hanging, termed as a free earlobe, whereas in some individuals, it is closely attached to the side of the head, referred to as an attached earlobe.

Chromosomes

• The chromosomes is a thread-like structure in the nucleus of a cell. It is formed of DNA and carries the genes.

The chromosome number is constant for the individuals of a species. For example, humans have 46 chromosomes.

Some more example are given in the following table.

		Number of chromosome
(i)	Mouse	40
(ii)	Gorilla	48
(iii)	Monkey	54
(iv)	Dog	78
(v)	Onion	16
(vi)	Maize	20

- A pair of corresponding chromosomes of the same shape, and size, one from each parent, is known as homologous chromosomes.

Sex Determination

- The process by which the sex of a person is determined is called sex determination.
- Sex chromosomes consist of two types: the X chromosome and the Y chromosome, denoted as XY.
- In females, the chromosomal composition include two X chromosomes, denoted as XX.



- If an X chromosome-carrying sperm fertilizes an egg, which also carries the X chromosome, the resulting combination of sex chromosomes will be XX, resulting in the birth of a female (girl).
- If a sperm carrying the Y chromosome fertilizes an egg, the resulting combination of sex chromosomes will be XY, leading to the birth of a male (boy).
- Thus, the male (father) is responsible for determining the gender of the baby.

Genes.

- Genes are specific DNA segment on a chromosome which determine hereditary characteristics.
- They play a role in passing traits from parents to their offspring.
- Genes work in pairs.
- Each genes possesses two alternative forms for a trait, which result in distinct effects, these alternate forms are known as alleles.



- The alternative forms of a character are known as traits.
- The gene which decides the appearance of an organism even in the presence of an alternative gene is known as dominant gene.
- The gene which decides the appearance of an organism only in the presence of another identical gene is called a recessive gene.
- The combination of genes present in an organism is called a genotype.
- Traits that are genetically determined and observable in an organism are referred to as its phenotype.
- In the trait of tongue rolling, individuals can exhibit one of two characters: they can either roll their tongue or not.

An example of inheritance: Tongue rolling.



The dominant gene for tongue rolling is denoted by 'R', while the recessive gene for non-rolling is represented by 'r'.

Therefore, in case of the tongue rolling phenotype, there are two type of genotype: homozygous dominant, consisting of a pair of identical alleles (RR) and heterozygous dominant, consisting of a pair of different allele (Rr).

Hollow symbol denote the standard expressed traits, specifically tongue rolling, while solid symbols indicate the atypical expressed traits namely non-rolling of the tongue.

In the provided chart, both the father and mother exhibit tongue rolling, among their children, two demonstrate tongue rolling while one does not possess this ability.

The presence of the recessive allele 'rr' in the third child.



indicating non-rolling, implies that the child inherited one r allele from each parent. Consequently, both parents must possess a heterozygous genotype for tongue rolling (Rr)

Sex-linked Inheritance

Sex-linked inheritance is the appearance of a trait which is due to the presence of an allele exclusively either on X chromosome or on the Y chromosome.

X-linked inheritance: Defect occur due to recessive genes, which are on the X chromosome.

For eg. Colourblindness and Haemophilia

• Haemophilia - In individuals with haemophilia, there is a risk of bleeding excessively or experiencing delayed clotting, potentially leading to fatal outcomes.

• Colourblindness: A person with colour blindness is unable to differentiate b/w red and green colours.

Cribs-cross inheritance.

87

- X-linked inheritance is also called cribs-cross inheritance.
- This is because the son may inherit a trait from the normal carrier method.

Y-linked Inheritance

Traits resulting from dominant genes located on the Y chromosome are exclusive to males. Examples of such traits include hypertrichosis (excessive hair growth on ears) and baldness.

Mendel's experiment-1 on Inheritance.

Gregor Mendel meticulously investigated the inheritance patterns, focus on the transmission of traits from one generation to the next. He is widely recognized as the father of Genetics.

- Mendel conducted breeding experiments on *Pisum Sativum* (garden pea). He selected the pea plant for three reasons:
 - There were many varieties available in alternative forms of a character.
 - Pure forms which bred true i.e. produced the same type generation after generation.
- The garden peas are self-pollinating but could be cross-pollinated artificially.
The following are the seven pairs of contrasting characters in the garden pea studies by Gregor Mendel.

Monohybrid Cross

- Mendel initially conducted crossbreeding experiments focusing on a single character.
- Example: Pure tall (TT) pea plant was crossed with dwarf (tt) plant.

• F_1 generation: All plants obtained were heterozygous tall. Plants from the F_1 generation were self-pollinated ~~due to~~ to obtain the F_2 generation.

F_2 generation: One homozygous tall plant, two heterozygous tall plants and one homozygous dwarf plant were obtained.

The ratio resulting from a cross involving two different traits of a single character is referred to as the monohybrid ratio.

Dihybrid Cross

Mendel carried out the cross breeding by taking two characters together.

Example: Plants with round yellow seed ($RRYY$) was crossed with the plants bearing wrinkled green seeds ($rryy$).

• F_1 generation: all plants obtained were heterozygous tall. Plants from the F_1 generation were self pollinated to obtain the F_2 generation.

• F_2 generation: One homozygous tall plant, two heterozygous tall plants and one homozygous dwarf plants were obtained.

• The phenotypic ratio was $9:3:3:1$

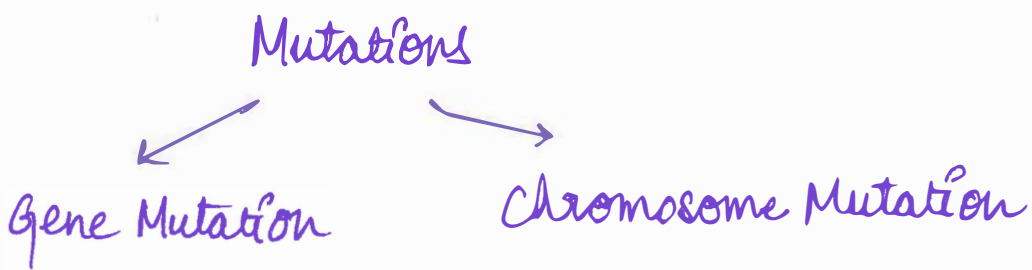
• The ratio obtained by breeding two pairs of contrasting characters is called dihybrid ratio.

Mendel laws of Inheritance.

1. Law of Dominance: Among a pair of contrasting characters coexisting together, only one is capable of expressing itself, while the other remains suppressed.
2. Law of Segregation: Two members of a pair of factors separate during the formation of gametes.
3. Law of Independent Assortment: In the presence of two pairs of contrasting characters, the distribution of one pair's member into gametes is independent of the distribution of the other pair.

Mutations.

Mutations refers to the abrupt alteration in one or more genes, or in the number or structure of chromosomes.



Sickle cell anaemia: It is a blood disease caused due to gene mutation. The mutation cause changes in DNA and result in the production of sickle-shaped RBCs.

Down's Syndrome - It is a mental deficiency caused due to one extra chromosome (chromosome no. 21)

Radioactive Radiations

Radioactive radiations alter the gene structure. The atomic bombing that took place during world war II in 1945 in Japan resulted in various deformities in the bodies of animal and plants.

Modern Applications of genetics.

Genetic engineering modifies an organism's genome by introducing new genes. This technique create genetically modified organisms (GMOs). One of its earliest applications was producing insulin.

