Chapter 52

Heredity & Variations

- **Genetics** is a branch of biology that deals with the **study** of **heredity** and **variations.**
- The term genetics was first used by **W. Bateson** (1905).
- **Gregor Johann Mendel** is called the **Father of Genetics**.
- **Archibald Garrod** is considered as the **Father of human genetics**.`

HEREDITY

- Heredity (like begets like) is the **study of transmission of characters from parents to offspring** or **from one generation to the next**.
- The characters that are passed from one generation to the other are called **hereditary characters**.
- This occurs through transfer of chromosomes from parents to offspring or individual to another. There is, therefore, **chromosomal basis of heredity**.
- The **character of heredity** are **fixed** for a particular organism.
- **Mendel used** the **term factors for the heredity unit which is now called as genes**.
- **Johannson** termed the word **phenotype** and **genotype** in addition to **genes**.
- The **physical basis of heredity** are **genes** while **chemical basis of heredity** is DNA.
- The prevailing view of heredity in pre-Mendelian era was **blending theory**. This theory states that **individual would represent the mixture of both the parents**.
- **Kolreuter** (1733-1806) for the first time obtained the experimental evidence that inherited traits tended to remain discrete, but he was unable to interpret them correctly.
- **R.de.Graaf (1641-1673)** suggested that **both the parent** should contribute to heredity.
- **Carl Nageli (1884)** was the first to propose a theory regarding heredity.
- **Davenport and Davenport (1910)** shows that skin colour in human is a polygenic trait and is due to atleast three separate genes.

Terms used in inheritance studies

- **Character** is a well defined morphological or physiological feature of an organism.
- **Trait** is the distinguishing feature of a character.
- **Gene** is the inherited factor that determine the biological character of an organism.
- A pair of contrasting characters is called **allelomorph or allele**.
- **Term allele** was **given by W. Bateson** for **alternative forms of same gene**, e.g., T and t, Y and y, R and r are pair of alleles.
- **Dominant allele** is one of the factor of an alleles pair which can express itself whether present in homozygous or heterozygous state, e.g., T (tallness in pea), R (round seed in pea).
- **Recessive allele** is the factor of an allele pair which is unable to express its effect in the presence of its contrasting factor in a heterozygote, e.g., t in Tt.
- The effect of **recessive factor** is **expressed only when it is present in the pure or homozygous state,** e.g., tt in dwarf pea plant.
- **Wild allele** is the one which was originally present in the population and is **dominant** and **widespread.**
- The **recessive allele** is **less common** and might have formed through mutation of wild allele. It is thus called **mutant allele**.
- The diploid condition in which the **alleles** at a **given locus** are **identical** is called **homozygous** or pure.
- In **homozygous condition**, organism **have two similar genes** or alleles for a particular character

in a homologous pair of chromosomes, e.g., **TT** or **tt**.

- Organisms containing **two different alleles** or individual containing both dominant and recessive genes of an allelic pair, e.g., **Tt**, is known as **heterozygous** or **hybrid**.
- When only one allelic pair is considered in cross breeding it is called **monohybrid cross**.
- When two allelic pairs are used for crossing it is called **dihybrid cross**.
- Involvement of more than two allelic pairs in a cross is called **polyhybrid cross**.
- Second stage of Mendel's experiment is called **F_1 generation** or **first filial generation**.
- Third stage of Mendel's experiment is called **second filial** or **F_2 generation**.
- **Complete penetrance** is 100% ability of an allelic combination to produce expected phenotype.
- **Incomplete penetrance** is failure of an allelic combination to provide cent percent phenotypic expression, e.g., polydactyly, diabetes mellitus.
- **Hybrid vigour** or **heterosis** is the superiority of hybrid over either of its parents in one or more traits.
- **Genotypes** is the sum total of heredity or genetic make up.
- **Phenotype** is the external feature of organism.
- When two different genotypes due to different environmental conditions give rise to same phenotype, then one is said to be **phenocopy of the other.**
- **Monohybrid ratio** is the ratio which is obtained in F_2 generation when a monohybrid cross is made and the offspring of F_1 generation are self-bred.
- **Dihybrid ratio** is a ratio which is obtained in the F_2 generation when a dihybrid cross is made and the offspring of F_1 generation are self-bred.
- **Trihybrid ratio** is the ratio obtained in F_2 generation raised from a trihybrid cross followed by selfing of F_1 individuals.
- When an individual is crossed with a parent it is called **back cross**.
- When an individual is crossed with the homozygous recessive parent it is called **test cross**.
- **Monohybrid test cross ratio** is 1 : 1 and **dihybrid test cross ratio** is 1 : 1 : 1 : 1.
- **Test cross is done for the following purposes –**
  - To prove that 2 types of gametes are produced by monohybrid, 4 types of gametes are produced by dihybrid and 8 types of gametes are produced by trihybrid.
  - That these gametes are produced in equal number.
  - Genotype of the offspring can be tested.
- If in one cross, individual X is used as male and Y as female and in the next cross Y is used as male and X as female, it is called **reciprocal cross**.
- The portion or region on chromosome representing a single gene is called **gene locus**.
- The alleles of a gene are present on the **same gene locus** on the homologous chromosomes.
- All the genotypes of all organisms in a population form the **gene pool**.
- **Pure line or pure breeding line** is a strain of individuals homozygous for all genes considered. The term was coined by **Johannsen**.
- **Punnet square** is a checker board which was devised by **R.C. Punnet** and used to show the result of a cross between two organisms.
- **Genome** is a complete set of chromosomes where every gene chromosome is represented singly as in a gamete.
- A single genome is present in **haploid cells**, two in **diploid cells** and many in **polyploid cells**.

Mendel's principles of inheritance (Mendelism)

- The first scientific study leading to formulations of law of inheritance was carried out by **Gregor Johann Mendel.**
- Mendel first represented his **rules of inheritance** in **1865**.
- Mendel was a monk in **Austria**.
- **Mendelian inheritance** or **Mendelian genetics** is a set of primary tenats that underlie much of genetics developed by G. Mendel in the latter part of 19th century.
- Mendel's laws of Heredity are described in his paper **"Experiments on Plant hybridization"** which was published in the fourth volume of **"Annual proceedings of Natural History Society of Brunn"**.
- Mendel's based his theory on experiments **involving cross pollination** between two plants or **self pollination** with a single plant.
- Mendel had **conducted hybridization experiments in garden pea**, *Pisum sativum*.

- Mendel's laws were rediscovered simultaneously by three great scientists namely **Huge de vries, Erich Von Tschermak** and **Carl correns**.
- The number of characters studied by Mendel in pea plant was **seven**.
- The number of chromosomes in *Pisum sativum* is **14 (2n).**
- Mendel restricted his experiments to one or few pairs of contrasting traits in each experiment.
- Self fertilization in pea can be prevented by removing anthers **(emasculation)** before pollen grains mature.
- **Mendel selected garden pea** for his experiment because –
  - It has a number of well defined **contrasting characters**.
  - It is having **bisexual flowers**.
  - It shows predominantly **self fertilisation** (autogamy).
  - **Hybridisation** or crossing is **easy** in pea.
  - It is having **short life span**.
- Mendel's experiment involved 4 steps as – **selection**, **hybridization**, **selfing** and **calculations**.
  - First he selected a true breeding variety.
  - Then he cross pollinated two contrasting true breeding varieties to get F_1 generation
  - Then he self pollinated F_1 offsprings to get F_2 and self pollinated F_2 to get F_3 generation.
- His results led to the formation of laws of genetics later.
- Mendel performed monohybrid and dihybrid crosses and gave **three principles** of **inheritance.**
- Mendel's **three principles** of **inheritance** are –
  - Law of dominance
  - Law of segregation or law of purity of gametes
  - Law of independent assortment.
- **Law of dominance** states that only one factor expresses itself in F_1 generation.
- F_2 expresses both the dominant and the hidden recessive factor in the ratio of **3 : 1** in the monohybrid cross.
- In a hybrid where both the contrasting alleles are present, only one factor/allele called **dominant** is able to **express** its effect while the other factor / allele called **recessive** remains **suppressed** in F_1 generation. This is called **Law of dominance**.
- **Exception to principle of dominance** are incomplete dominance and codominance.
- **Incomplete dominance** is the phenomenon where dominant allele do not completely express itself.
- Incomplete dominance is not blending inheritance because **parental characters reappear in F_2 generation**.
- This phenomenon was first studied in flower colour of *Mirabilis jalapa* or **four O'clock plant**.
- The phenotypic as well as genotypic monohybrid ratio in F_2 generation in incomplete dominance is **1 : 2 : 1** *i.e.*, pure dominant : hybrid : pure recessive.
- F_1 generation expresses a phenotype which is **intermediate** between those of the parent.
- When blue Andalusian are crossed among themselves, they produce splashed white, blue and black offspring in the ratio of **1 : 2 : 1 due to incomplete dominance.**
- **Overdominance** is another dominance relationship in which the phenotype of heterozygote is not equal to that of either homozygote.
- Heterozygote with overdominance alleles have a phenotype more extreme than either homozygote.
- In codominance, both allelic genes of a genetic trait are equally expressive *i.e.*, the dominant character is not able to suppress the recessive character and thus both the characters appear side by side in F_1 hybrids.
- **Principle of segregation** states that, "**when a pair of contrasting factor or gene are brought together in a hybrid; these factors do not blend or mix up but simply associate themselves and remain together and separate at the time of gamete formation**".
- This law can **also be stated as** alleles pairs segregate during gamete formation and the paired condition is restored by random fusion of gametes during fertilization.
- The above law is also known as "**Law of purity of gametes**" because each gamete is pure in itself *i.e.*, having either T (*i.e.*, gene for tallness) or t (*i.e.*, gene for dwarfness).
- Mendel formulated this law **with the help of monohybrid cross**.
- The third principle of heredity is called the **principle of independent assortment**.
- It states that the genes of different characters located in different pairs of chromosomes are **independent of one another in their segregation during gamete formation**.

- Independent assortment is not applicable for the genes located on the same chromosome, *i.e.*, linked genes.
- Linkage is an **exception of Mendelian principles** because characters studied in pea plant by him were located on different chromosomes or the distance separating the syntenic loci was sufficiently great so that the genes were inherited as though they were on separate chromosomes.
- The genes controlling the seven pea characters studied by Mendel are now known to be located on **four chromosomes (1, 4, 5, 7)**.
- Mendelian recombinations were mainly **due to independent assortment.**
- Test cross is also **applicable to dihybrid cross.**
- A dihybrid test cross give a **1 : 1 : 1 : 1 ratio**, indicating that two pairs of factors are segregating and assorting independently.
- **Atavism** is the phenomenon during inheritance wherein a character remains hidden for several generation and then suddenly gets expressed unchanged in one or more individuals.
- Monohybrid phenotypic ratio is **3 : 1**.
- Monohybrid genotypic ratio is **1 : 2 : 1**.
- Dihybrid genotypic ratio is **1 : 2 : 1 : 2 : 4 : 2 : 1 : 2 : 1**
- Dihybrid phenotypic ratio is **9 : 3 : 3 : 1**
- **Importance of Mendelism** are –
  - On the basis of Mendelism, different breeds in animals and varieties of plants have been produced.
  - Science of **eugenics** (development of superior progeny) is based on Mendelism.
  - On the basis of Mendelism, **heterosis** has been utilised in different organisms.
- Mendel was **successful in his experiments with pea plant** because
  - He selected pure breeding, one or two characters at one time for his breeding experiments.
  - He took those traits which did not show linkage, interaction or incomplete dominance.
  - He used statistical methods and law of probability analysing his results.
  - Pea had contrasting expression of traits.
  - Pea can be cross bred manually but itself undergoes self breeding.
  - There is little chance of pollen contamination.

Gene interaction

- Gene interaction is **the modification of normal phenotypic expression of genes due to their alleles and non allelic genes**.
- Gene interaction is a **post mendelian discovery**.
- Gene interaction is of **two types** – intragenic and intergenic.
- In **intragenic interaction**, two alleles of a gene which are present on the same gene locus on the two homologous chromosomes, react to produce modified phenotype. Eg., incomplete dominance, codominance, multiple alleles and lethal genes.
- **Intergenic interaction** is one where two or more independent genes belonging to same or different chromosomes interact to form a different expression.
- Intergenic interaction includes epistasis, duplicate genes, complementary, genes, supplementary genes, inhibitory genes etc.
- **Modified Mendelian ratio** are expressed as 9 : 7, 9 : 3 : 4, 12 : 3 : 1, 15 : 1, 13 : 3.

Incomplete dominance

- The **main objection to the Mendel's principle** of genetics **was incomplete dominance**.
- **Incomplete dominance** is the phenomenon where dominant allele do not completely express itself.
- **Correns** discovered **incomplete dominance** and **cytoplasmic inheritance**.
- Incomplete dominance is not blending inheritance because **parental characters reappear in F_2 generation**.
- This phenomenon was first studied in flower colour of *Mirabilis jalapa* or four O'clock plant.
- The phenotypic as well as genotypic monohybrid ratio in F_2 generation in incomplete dominance is **1 : 2 : 1** *i.e.*, **pure dominant : hybrid : pure recessive**.
- F_1 generation expresses a phenotype which is **intermediate** between those of the parent. Eg pink flowers are obtained when red and white flowers plants are crossed.

Codominance

- Codominance is the **phenomenon of two alleles lacking dominant recessive relationship** and both express themselves in the organisms.
- The **codominant alleles** are able to express

themselves independently when present together and co-dominance has been reported in roan characters of cattle.

- The codominant alleles are shown with same capital letter but with different superscripts like I^AI^B for allele in human blood group AB and Hb^A Hb^S for normal and sickle celled erythrocytes.

Multiple alleles

- **More than two alternate forms of a gene** present on the same locus are called **multiple alleles**.
- There is **absence of crossing over** in multiple alleles and the mode of inheritance in case of multiple alleles is called **multiple allelism.**
- The well known example of multiple allele in human is **blood group**, which also shows codominance.
- **Landsteiner** discovered the three blood groups in man (**A, B** and **O**).
- Blood group **AB** was discovered by **de Castello and Steini (1902)**.
- There are more than two alleles of the same gene e.g. 15 alleles for eye colour in *Drosophila*.
- Multiple alleles express different alternatives of the same characters.
- Different alleles show codominance, dominance-recessiveness or intermediate dominance amongst themselves.
- They however, follow Mendelian pattern of inheritance.

Lethal genes

- A lethal gene can be defined as **a gene whose phenotypic effect is sufficiently drastic to kill the bearer**.
- Lethal genes **control some vital functions of the organism and cause death of the organisms** in **pure recessive** or **pure dominant form**.
- Lethal gene were **first discovered by Cuenot**.
- In **absolute lethality** individual dies in embryonic stage. For eg. yellow fur in mice.
- In **sublethality** individual dies before reproductive maturity. For eg. sickle cell anaemia.
- In **delayed lethality** individual dies after sexual maturity.
- **Albinism** in corn is **due to lethal genes**.
- **Inhibitor** or **suppressor genes** are non-lethal gene **without any expression of its own but suppress or inhibit the expression of a non-allelic gene**.
- For example, in Rice I-gene inhibits the expression of dominant purple colour gene (P) so that the leaves are green in its presence (I-P-). Green leaves also occur when the leaf colour gene is recessive (*iipp*). A cross between IIPP and *iipp* (both green) yields hybrid greens (I*i*P*p*) which on self breeding form **3 purple to 13 green plants**.

Epistasis

- Epistasis can be defined as the **phenomenon of gene interaction whereby one gene interferes with the phenotypic expression of another non allelic gene or genes**.
- The gene or locus which suppresses or masks the action of a gene at another locus is called **epistatic gene.**
- The gene or locus whose expression is suppressed by an epistatic gene is called **hypostatic gene**.
- **Hypostastasis** is the phenomenon by which the effect of a gene gets suppressed due to the presence of a non allelic gene.
- An **epistatic hypostatic relationship** between two loci is **similar** to a dominant recessive relationship between alleles at a particular loci.
- A **dominant epistatic allele** suppress the expression of a non allelic gene, the latter may be dominant or recessive.
- The dihybrid ratio for dominant epistasis is **12 : 3 : 1**.
- In **recessive epistasis**, epistatic gene suppresses the expression of non-allelic gene only when it is in homozygous recessive state.
- Recessive epistasis or supplementary gene ratio is **9 : 3 : 4 (dihybrid ratio)**.
- **13 : 3** is dominant recessive epistasis ratio.
- In a cross between black (CCaa) and albino (AAcc) guinea pig F_2 exhibits **9 agouti : 3 black : 4 albino**.

Complementary, duplicate, supplementary and polymetric genes

- If two genes present on different loci produce the same effect when present alone but interact to form a new trait when present together are called **complementary genes**.
- **Bateson and Punnet (1906)** observe complementary gene in *Lathyrus odoratus*.
- There are two white varieties of Sweet pea (*Lathyrus odoratus*) controlled independentally by two different genes (C-pp and ccP).

Table : Expected ratios in different types of crosses with examples

	Characters	Expected ratios	Example
1.	**Monohybrid cross**	Phenotypic : 3 : 1	*Pisum sativum*
		Genotypic : 3 : 1	*Pisum sativum*
2.	**Dihybrid cross**	Phenotypic : 9 : 3 : 3 : 1	*Pisum sativum*
3.	**Incomplete dominance**	1 : 2 : 1	*Mirabilis jalapa*
4.	**Complementary genes**	9 : 7	*Lathyrus odoratus*
5.	**Supplementary genes**	9 : 3 : 4	Coat colour in mice
6.	**Modified supplementary genes (collaboration)**	9 : 3 : 3 : 1	Poultry birds – comb pattern
7.	**Dominant epistasis**	12 : 3 : 1	Fruit colour in *Cucurbita*
8.	**Recessive epistasis**	9 : 3 : 4	Coat colour in mice
9.	**Duplicate genes**	15 : 1	Fruit shape in *Capsella bursa-pastoris*
10.	**Polymeric gene (duplicate genes with cumulative effect)**	9 : 6 : 1	*Cucurbita pepo*
11.	**Supressor gene (Inhibitor gene)**	13 : 3	Leaf colour in rice

- Dominant gene C produces an enzyme that converts the raw material for flower pigmentation into chromagen. Dominant gene P produces another enzyme that oxidises chromagen into purple coloured anthocyanin. Therefore, the dominant alleles of both the genes are required for expression of flower colour.
- **Complementary gene ratio** is **9 : 7**.
- There are two (or more) independent genes found on different chromosomes which produce the same or nearly similar phenotypic effect in the dominant state, producing same intensity of effect even when present together, so that dominant phenotype is more abundant. Such genes are called **duplicate genes** or **pseudoalleles**.
- **Duplicate dominant gene ratio** is **15 : 1**.
- **Supplementary genes** are two non-allelic genes in which one type of gene produces its effect whether the other is present or not and the second (supplementary) gene produces its effect only in the presence of the first usually forming a new trait.
- F_2 dihybrid ratio is 9 : 3 : 4.
- **Polymeric genes** are duplicate genes with cumulative/additive effect, *i.e.*, two independent dominant genes (whether homozygous or heterozygous) having similar phenotypic affect individually but produce a new cumulative effect (similar in homozygous and heterozygous states) when present together.
- **9 : 6 : 1** is the ratio of **duplicate genes** with **cumulative effect**.

Pleiotropic genes

- When a **gene affects** many aspects of phenotype or **controls several phenotypes**, it is said to be **pleiotropic genes** and this phenomenon is called **pleiotropy**.
- When a number of related changes are caused by a pleiotropic gene, the phenomenon is called **syndrome**.
- Pleiotropy is expressed by **sickle cell anaemia, haemophilia**, etc.
- Sickle cell anaemia disease is caused when a gene responsible for haemoglobin produced by recessive alleles differs in **one amino acid**. It incorporates valine in place of **glutamic acid**.
- In human beings, **Marfan's syndrome**, characterised by long limbs, slender body, hypermobility of joints, lens dislocation and susceptibility to cardiac diseases are caused by **single pleiotropic gene**.
- **Galton** coined the term **eugenics**.

Pedigree analysis

- A record of the occurrence of a trait in several

generation of a human family is called **Pedigree analysis**.

- **Male member** in pedigree are shown by square □ and **female** by circle O.
- Siblings are represented **horizontally on a line** in order of birth.
- **Solid symbols** (●) represent the traits being invested and **open symbols** O or □ represent the normal traits.
- Pedigree analysis **helps** in identifying the inheritance of specific traits and their possibility of showing up in the offsprings. It is of great significance in the study of genetics.

Quantitative (polygenic) and qualitative inheritance

- **Quantitative or polygenic inheritance** is that type of inheritance in which the complete expression of a trait is controlled by two or more genes in which a dominant allele of each gene contributes only a unit fraction of a trait and the **total phenotypic expression is the sum total/additive/cumulative effect of all the dominant alleles of genes/ polygenes**.
- In quantitative inheritance traits are **expressed in continuous fashion**.
- The genes involved in quantitative inheritance are called **polygenes** or **cumulative genes.**
- A **polygene** is defined as a gene where a dominant allele controls only a unit or partial quantitative expression of a trait.
- **Nilsson-Ehle (1908)** was first to experimentally prove quantitative inheritance.
- Polygenic inheritance is also called **quantitative inheritance** since so many grades between two extremes appear, eg. skin colour in human, intelligence etc.
- **Davenport and Davenport (1910)** shows that skin colour in human is a polygenic trait and is due to at least three separate genes.
- In quantitative inheritance **traits are expressed in continuous fashion**.
- **Qualitative inheritance** is that **type of inheritance in which one dominant allele influences the complete trait so that two such alleles do not change the phenotype**.
- **Monogene** is a gene in which one dominant allele controls the complete or qualitative expression of a trait.

VARIATIONS

- The characteristic differences exhibited by the individual of the same species, race and family is called **variations**.
- Variation caused due to genetic difference are called **genetic variation** or **heredity variations**.
- The variation caused due to environmental factors and which is not fixed is called **environmental variation**.
- Heredity variations are **transmitted from generation to generation** whereas environmental variation are **temporary and do not relate with last or next generation**.
- Heredity variation within a progeny results due to **sexual reproduction**.
- **Somatic variations** affect the somatic or body cells of the organisms and these die with the death of the individual and thus are **non inheritable**.
- Somatic variations are also called **modifications** of **acquired characters** because they are acquired by an individual during its life time.
- Somatic variations are **caused by three factors** namely **environment**, **use and disuse of organs** and **conscious efforts**.
- **Continuous variations** are **fluctuating variations** which oscillate around a mean or average of the race, variety and species.
- Continuous variations are also called **recombinations** because they are formed due to recombination of alleles caused by –
  - Chance separation/segregation of chromosomes at the time of meiosis (sporogenesis in plants and gametogenesis in animals).
  - Crossing over or exchange of chromatid segments during meiosis.
  - Chance aggregation of chromosomes during fertilization.
- Continuous variations are of **two types** –
  - **Meristic**, influencing number of parts like number of grains in an ear of wheat, number of tentacles in *Hydra*.
  - **Substantive**, influencing appearance like height, colour, yield of milk or eggs.

- **Discontinuous variations** are **mutations** which are sudden, unpredictable inheritable variations not connected with the average by any intermediate stages.
- Discontinuous variations are **source of all germinal variations and most of evolution**.
- Discontinuous variations are **caused by chromosomal aberrations, change in chromosome number and gene mutations**. Depending upon the effect, they are of two types –
  - **Indeterminate variations**, which occur in any conceivable direction and to any degree.
  - **Determinate variations**, which are variations in particular direction with selective or adaptive importance and are also called **orthogenic variations**.
- Variations make some individual better fitted in the struggle for existence.
- They help the individuals to adapt themselves according to the changing environment.
- Discontinous variations produce new traits in the organisms.
- Variations allow breeders to improve races of useful plants and animals for increased resistance, better yield, quicker growth and lesser input.
- They constitute the raw material for evolution.
- Variations gives each organism a distinct individuality.
- Because of variations species do not remain static. Instead, they are slowly getting modified forming new species with time.
- Pre-adaptations caused by the presence of neutral variations are extremely useful for survival against sudden changes in environment, e.g., resistance against a new pesticide or antibiotic.

End of the Chapter