CHAPTER-4

PRINCIPLES OF INHERITANCE AND VARIATION

Topic-1

Mendel's laws and Chromosomal Theory

Concepts Covered ● *Heredity, variation, Mendel's laws of Inheritance, Non-Mendelian inheritance, Chromosomal theory, Linkage and Recombination.*

Revision Notes

- **Heredity (L. hereditas - heirship or inheritance) :** It is the sum of all biological processes by which particular characteristics are passed on from parents to their offspring, either through asexual or sexual reproduction.
- **Variation:** Tendency of differences in various traits of individuals of a progeny from one another and their parents.

Mendel's Laws of Inheritance :

- **Hybridization Experiments on Garden Pea (*Pisum sativum*)**
  - Mendel selected 7 pairs of contrasting traits of true breeding pea varieties.

S. No.	Characters	Dominant	Recessive
1.	**Height of the stem**	Tall (T)	Dwarf (t)
2.	**Colour of the flower**	Violet/ Red (R)	White (r)
3.	**Position of the flower**	Axial (A)	Terminal (a)
4.	**Shape of pod**	Full/ Inflated (I)	Constricted (i)
5.	**Colour of pod**	Green (G)	Yellow (g)
6.	**Shape of seed**	Round (R)	Wrinkled (r)
7.	**Colour of seed/cotyledons**	Yellow (Y)	Green (y)

Inheritance of One Gene

- **Monohybrid Cross :**
  - A cross involving two plants differing in one pair of contrasting characters.
  - e.g., Mendel crossed tall and dwarf pea plants to study the inheritance of one gene.
- **Steps in Making a Cross of Pea :**
  - Selection of two pea plants with contrasting characters.
  - Removal of anthers (emasculation) of one plant to avoid self- pollination. This is a female parent now.
  - Collection of pollen grains from the other plant (male parent) and transfer to female parent for pollination.
  - Collection of seeds and production of offspring.
  - Mendel made similar observations for other pairs of traits and proposed that factors were inherited from parent to offspring. Later, these factors were called genes.
  - The **F_1 generation** (Tt) when self-pollinated, produces gametes T and t in equal proportion.
  - Mendel self-pollinated the **F_2 generation** plants.
  - He found that dwarf F_2 plants continued to generate dwarf plants in F_3 & F_4 generation.
  - He concluded that the **genotype** of the dwarf was homozygous- ***tt*.**
- **Monohybrid Phenotypic Ratio :** 3 Tall : 1 Dwarf = 3 : 1
- **Monohybrid Genotypic Ratio :**

  1 Homozygous tall (TT) : 2 Heterozygous tall (Tt) : 1 Homozygous dwarf (tt)
- **Back cross and Test cross**
  - **Back cross :** Crossing of F_1 hybrid with either of its parent.
  - **Test cross :** Crossing of an F_1 hybrid with its recessive parent (Test cross ratio=1:1). It is used to find out the unknown genotype. Mendel conducted a test cross to determine the F_2 genotype.

Mendel's Principles or Laws of Inheritance :

1. **Principle of Dominance**
   - Characters are controlled by discrete units called factors.
   - Factors occur in pairs.
   - In a dissimilar pair of factors or contrasting **alleles** i.e., in heterozygous condition, only one member of the pair expresses its effect in the hybrid and is called dominant while the manifestation of the other is masked and is called recessive.
2. **Law of Segregation**

   This law states that allelic pairs separate or segregate during gamete formation and randomly unite at fertilization, thus homozygous parent produces similar gametes. Heterozygous parent produces two kinds of gametes, each having one allele in equal proportion.

   The Concept of Dominance
   - In heterozygotes, there are dominant and recessive alleles.
   - The normal (unmodified or functioning) allele of a gene produces a normal enzyme that is needed for the transformation of a substrate.
   - The modified allele is responsible for the production of
     (i) The normal/less efficient enzyme or
     (ii) A non-functional enzyme or
     (iii) No enzyme at all
   - **In the first case :** The modified allele will produce the same phenotype like the unmodified allele. It becomes dominant.
   - **In 2nd and 3rd cases :** The **phenotype** is dependent only on the functioning of the unmodified allele. Here, the modified allele becomes recessive.

Key Words

Alleles or allelomorphs : A pair of Mendelian factors or genes located on the same locus of two homologous chromosomes of an individual which control the expression of a trait or character are called alleles or allelomorphs.

F_1 generation: Hybrids Produced from a cross between the genetically different individuals called parents. e.g., Tt individuals are produced in F_1 generation from a cross between TT and tt parents.

F_2 generation : It is the generation of individuals which arises as a result of interbreeding or selfing amongst individuals of F_1 generation.

Genotype : (Gk. Geno-race; typos — image). It is the genetic constitution of individual with regard to one or more characters irrespective that whether the genes are expressed or not, for e.g., genotype of hybrid tall pea plant is Tt, pure tall TT and pure dwarf tt.

Phenotype : (Gk. Pheno — to appear, typos — image): It is observable or measurable distinctive structural or functional characteristic of an individual. e.g., phenotypic tall pea plant can be genotypically TT or Tt.

Non-Mendelian Inheritance

(a) Incomplete Dominance

- It is an inheritance in which heterozygous offspring shows an intermediate character between two parental characteristics. e.g., Flower colour in Snapdragon (dog flower or *Antirrhinum* sp.) and *Mirabilis jalapa* (4'O clock plant).
- Here, phenotypic and genotypic ratios are the same.
- Phenotypic ratio = 1 Red : 2 Pink : 1 White
- Genotypic ratio = 1 (RR) : 2 (Rr) : 1(rr)
- This means that R was not completely dominant over r.

Key Fact

Mendel, besides pea plant, also worked on Hawkweed (*Hieracium*) and lablab. But he failed to obtain same results as he found availability and in garden pea due to parthenogenesis and non- availability of pureline seeds.

(b) Co-dominance

- It is the inheritance in which both alleles of a gene are expressed equally and independently in a hybrid i.e., both the alleles are dominant e.g., ABO blood grouping in humans.
- ABO blood groups are controlled by the gene.
- The gene (*I*) has three alleles I^A, I^B and *i.* However, a person can have any two of these three alleles. I^A and I^B both are dominant alleles while *i* is a recessive allele.
- The alleles I^A and I^B produce antigen A and antigen B respectively on the RBC surface while allele *i* doesn't produce any antigen.
- When I^A and I^B are present together they both express their types of surface antigen A and B. This is due to co-dominance.

(c) Multiple Allelism

- Here, more than two alleles govern the same character.
- Since in an individual, only two alleles are present, multiple alleles can be found only when population studies are made e.g., ABO blood grouping (3 alleles : I^A, I^B & i). The skin colour and height of humans are also examples of multiple alleles.

(d) Pleiotropy

- Pleiotropy is the phenomenon in which one gene controls many traits. For example, the gene in pea plants that controls the round and wrinkled texture of seeds also influences the phenotypic expression of starch grain size.
- So, if the starch grain size is considered as the phenotype, then from this angle, the alleles show incomplete dominance.
- Therefore, dominance is not an autonomous feature of a gene or the product that it has information for. It depends as much on the gene product and the production of a particular phenotype.

Inheritance of Two Genes

Dihybrid Cross

- A cross between two parents differing in two pairs of contrasting characters.
- Mendel made some dihybrid crosses e.g., Cross between the pea plants with round shaped and yellow coloured seeds (RRYY) and wrinkled shaped and green coloured seeds (rryy).
- On observing the F_2 generation, Mendel found that the yellow and green colour segregated in a 3:1 ratio.
- Round and wrinkled seed shape also segregated in a 3:1 ratio.

- Thus, the segregation of one pair of contrasting characters (Round and wrinkled shape) is independent of the segregation of another pair of contrasting character (yellow and green) colour and also that some new combinations of character appear in F_2 generation as the alleles get randomly rearranged in the offsprings at the time of fertilization.
- Dihybrid genotypic ratio: 1 : 2 : 2 : 4 : 1 : 2 : 1 : 2 : 1
  RRYY =1; RRYy =2; RrYY = 2; RrYy = 4; RRyy = 1; Rryy = 2; rrYY = 1; rrYy = 2; rryy = 1
- **Dihybrid Phenotypic ratio :**
  Round yellow 9 : Round green 3 : Wrinkled yellow 3 : Wrinkled green : 1, i.e., 9 : 3 : 3 : 1
  The ratio 9 : 3 : 3 : 1 can be derived as a combination series of 3 yellow : 1 green, with 3 round : 1 wrinkled. i.e.,
  (3 : 1) (3 : 1) = 9 : 3 : 3 : 1

3. Mendel's Law of Independent Assortment :

- It states that when more than one pair of characters are involved in a cross, the segregation of one pair of contrasting characters is independent of the segregation of other pair of contrasting characters and also that new recombinations of characters along with the parental type also appear in the F_2 generation.

Non-recognition of Mendel's work

- **Mendel's work remained unrecognizable till 1900 because :**
  - **(a)** Communication was not easy.
  - **(b)** Non-recognition of Mendel as a scientist.
  - **(c)** His mathematical approach was new and unacceptable.
  - **(d)** He used statistical calculations which were beyond the comprehension of the biologists of his time.
  - **(e)** Chromosomes, mitosis and meiosis were not known in Mendel's time.
  - **(f)** The concept of genes (factors) as stable and discrete unit was not accepted. Mendel could not explain the continuous variations seen in nature.
  - **(g)** Mendel could not provide any physical proof for the existence of factors.
- In 1900, de Vries of Holland, Correns of Germany & Von Tschermak of Austria independently rediscovered Mendel's results and proclaimed their conclusions as Mendel's Laws of inheritance.

Chromosomal Theory (1902)

- The Chromosomal theory was proposed independently by Walter Sutton and Theodore Boveri in 1902.
- Walter Sutton & Theodore Boveri proposed that the pairing and separation of a pair of chromosomes during meiosis lead to the segregation of pair of factors.
- Sutton united chromosomal segregation with Mendelian principles and called it the Chromosomal Theory of Inheritance.
- **It states that :**
  - **(a)** Chromosomes are vehicles of heredity i.e., they are transmitted from parents to offspring.
  - **(b)** Two identical chromosomes form a homologous pair. Genes are present in a linear fashion on chromosomes.
  - **(c)** They segregate at the time of gamete formation.
  - **(d)** Independent pairs segregate independently of each other.
  - **(e)** Chromosomes are mutable.
  - **(f)** Sex chromosomes determine the sex of an individual.

Parallelism between Genes (Mendelian factors) & Chromosomes :

- Mendelian factors as well as chromosomes are transferred from generation to generation.
- The chromosomes occurs in homologous pairs. The genes also occurs in pairs (allelic pairs).
- Both chromosomes and genes segregate at the time of gamete formation in such a way that gametes receive only one chromosome & similarly one allele of each pair.
- Different pairs of chromosomes segregate independently of each other. Similarly, one pair of alleles segregates independently of another pair.
- Fusion of two (male & female) gametes brings about the diploid chromosome number as well as the allelic pairs in the offsprings.
- **Thomas Hunt Morgan** proved the Chromosomal Theory of Inheritance using fruit flies **(***Drosophila melanogaster***).**
  He took fruit flies as a suitable material because :
  - **(a)** It breeds very quickly.
  - **(b)** Short generation time (life cycle : 12-14 days).
  - **(c)** Breeding can be done throughout the year.
  - **(d)** Hundreds of progenies are produced per mating.
  - **(e)** They can grow on a simple synthetic medium.
  - **(f)** Male and female flies are easily distinguishable.

Linkage and Recombination

- **Recombination :** It is a process by which pieces of DNA are broken and recombined to produce a new combination of alleles.

- **Linkage :** Physical association of two or more genes on a chromosome, which show the tendency to inherit together. They do not show independent assortment.
- Morgan et. al crossed yellow body and white eyed females with wild type brown body and red-eyed males and inter-crossed F_1 offsprings. He found that the two genes did not segregate independently, resulted in deviation from normal dihybrid ratio 9 : 3 : 3 : 1 in F_2 generation because the appearance of parental combinations were higher than the non-parental and new recombinations.
- Morgan further carried out several dihybrid test crosses in *Drosophila* to study sex-linked genes.
  Cross A : Double recessive, yellow-bodied, white-eyed females (yw/yw) X hybrid brown-bodied, red-eyed males (Y'W'/YW) (wild type).
  Cross B : Double recessive, white-eyed, miniature winged (wm/wm) X hybrid red eyed, large winged (W'm/Wm) (wild type).
- **Morgan in the above crosses found that :**
  (a) The two genes did not segregate independently of each other and the F_2 ratio deviated from the 9 : 3 : 3 : 1 ratio.
  (b) Genes were located on the X chromosome.
  (c) When two genes were situated on the same chromosome, the proportion of parental gene combinations was much higher than the non-parental type. This is due to linkage.
  (d) Genes for white and yellow were very tightly linked and showed only 1.3% new recombination while white and miniature wings showed 37.2% recombination (loosely linked).
  (e) Tightly linked genes show low recombination.
  (f) Loosely linked genes show high recombination.
  The strength of linkage is inversely proportional to the distance between two linked genes. Thus, the linkage between y & w alleles is stronger than the linkage between w & m alleles.
- **Linkage groups :** All the genes present together on a single chromosome make up a linkage group. The total number of linkage groups in an organism is equal to its haploid number of chromosomes or number of homologous pairs in diploid organisms.
- Alfred Sturtevant used the recombination frequency between gene pairs as a measure of the distance between genes and 'mapped' their position on the chromosome.
- Recombination frequency or the cross over value (COV) can be calculated by the following formula.

$$\text{COV} = \frac{\text{Number of recombinants}}{\text{Total number of offsprings}} \times 100$$

- Genetic maps are used as a starting point in the sequencing of genomes as was done in Human Genome Project.

Mnemonics

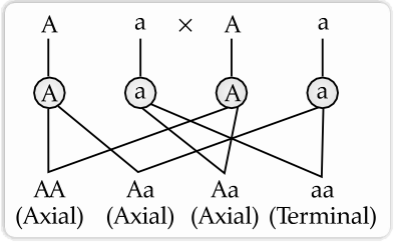
Concept: Non-Mendelian Inheritance
Mnemonics: I Care **M**endel's **P**rinciples**.**

Interpretations: Incomplete dominance, **C**o-dominance, **M**ultiple alleles, **P**leiotropy

Example 1

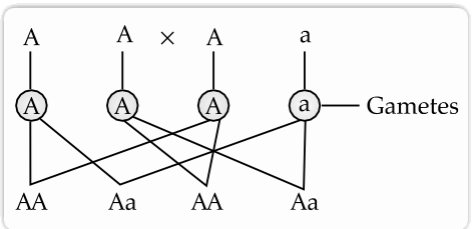
Q. In a garden pea plant, the flowers may be axial (A) position. Find out the proportion of terminal (a) in position. Find out the proportion the offspring in the following crosses would be expected to be terminal in position.
(i) Aa ×Aa
(ii) AA ×Aa

Sol. (i)



25 percent of the offspring are terminal in position

(ii)



None of the offspring is terminal in position

Topic-2 Sex Determination and Chromosomal Disorder

Concepts Covered • *Sex determination, Mendelian disorders, chromosomal disorders.*

Revision Notes

- **Sex determination**
  - The method by which the distinction between male and female is established in a species is called sex determination.
  - Sex of an individual is finalized at the time of zygote formation.
- **Autosomes and Sex chromosomes (allosomes)**
  - Autosomes are chromosomes other than sex chromosomes. They contain genes that determine somatic characteristics.
  - Number of autosomes is the same in males and females.
  - Sex chromosomes (X & Y) are the chromosomes that are involved in sex determination.
  - **Henking (1891)** studied spermatogenesis in some insects and observed that 50 % of sperm received a nuclear structure after spermatogenesis, whereas the other 50 % of sperms did not receive it.
  - Henking called this structure as the **X body** (later it was called as **X-chromosome**).
- **Mechanism of Sex Determination**

(i) Chromosomal sex determination : It is based on heterogamety i.e., the occurrence of two types of gametes in one of the two sexes. It is of the following types :

(a) XX-XO mechanism :

Here, the male is **heterogametic** i.e., XO besides autosomes (gametes with X and gametes without X) and female is **homogametic** i.e., XX (all gametes are with X chromosomes).

(b) XX-XY mechanism :

Male is heterogametic (X & Y) and female is homogametic (X only). e.g., Human and *Drosophila.*

(c) ZZ-ZW mechanism :

Male is homogametic (ZZ) and female is heterogametic (Z & W). e.g., Birds.

(d) ZO-ZZ mechanism : Females have only Z-chromosomes besides autosomes and males have a pair of Z-chromosomes e.g., in cockroaches.

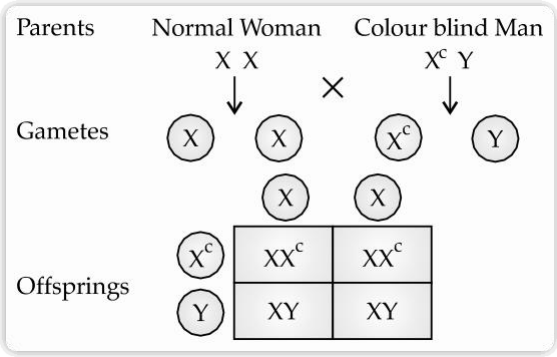
XX-XO & XX-XY mechanisms show male heterogamety. ZZ-ZW mechanism shows female heterogamety. Females have only Z chromosome besides autosomes and males have a pair of Z chromosome as seen in cockroaches.

Example 2

Q. Show the process of sex-linked inheritance of colour blindness

(a) If a colour-blind man (X^cY) marries a girl with normal vision (XX). Show the possibility of normal boy and carrier girl.

Sol.



Result shows that 50 percent offspring are girls and they are carrier and 50 percent offspring are boys but they are normal.

- **Sex Determination in Humans (XX-XY type)**
  - Human has 23 pairs of chromosomes (22 pairs are autosomes and 1 pair is sex chromosome).

- A pair of X-chromosome (XX) is present in the female, whereas the X and Y chromosome are present in male.
- During spermatogenesis, males produce 2 types of gametes i.e., 50 % with X-chromosome and 50 % with Y-chromosome.
- Females produce the only ovum with X-chromosome.
- There is an equal probability of fertilization of the ovum with the sperm carrying either the X or Y chromosome.
- The sperm determines whether the offspring will be male or female.

(ii) Environmental Sex-determination : Determination of sex depends upon the environmental condition. Environmental factors like temperature, etc., determine whether the zygote will develop into male or female. e.g., In turtles and crocodile.

(iii) Genetic balance mechanism of sex determination : The sex of the individual is decided by the ratio of X-chromosome and autosome, as it is found in ***Drosophila***.

(iv) Cytoplasmic Sex-determination : Cytoplasmic or fertility factor called as an F^+ factor located in plasmid determines the sex as it is found in some bacteria.

Genetic Disorders

- There are two types of genetic disorders namely, Mendelian disorders and Chromosomal disorders.

(1) Mendelian Disorders

- It is caused by alteration or mutation in a single gene.
- The pattern of inheritance of Mendelian disorders can be traced in a family by the pedigree analysis. e.g., Haemophilia, Cystic fibrosis, Sickle-cell anaemia, Colour blindness, Phenylketonuria, Thalassaemia, etc.
- Mendelian disorders may be dominant or recessive.

Mnemonics

Concept: Mendelian Disorders
Mnemonics: **H**ighlight **T**he **C**oncepts **C**learly

Interpretations: Haemophilia, Thalassaemia, Cystic fibrosis, Colour blindness**.**

(a) Colour blindness

- It is a recessive sex-linked trait in which the eye fails to distinguish red and green colours.
- The normal gene and its recessive allele are carried by X-chromosome.
- In female, colour blindness appear when both the sex chromosomes carry the recessive gene.

(b) Haemophilia (Royal disease)

- Sex-linked recessive disease.
- In this, a protein involved in the blood clotting is affected.
- A simple cut results in non-stop bleeding.
- The heterozygous female (carrier) for haemophilia may transmit the disease to sons.
- The possibility of a female becoming a haemophilic is very rare because mother has to be at least carrier and the father should be haemophilic (inviable in the later stage of life).
- Queen Victoria was the carrier of this disease. So her family pedigree shows many haemophilic descendents.

(2) Chromosomal Disorders

- They are caused due to the absence or excess or abnormal arrangement of one or more chromosomes.
- These are of two types namely,
  (a) Aneuploidy **(b)** Euploidy.

(a) Aneuploidy

- The gain or loss of chromosomes due to failure of segregation of chromatids during cell division.
  It includes,
  (a) Nullisomy (2n-2) : A complete homologous pair is lost from diploid set.
  (b) Monosomy (2n-1) : One chromosome is lost from the diploid set.
  (c) Trisomy (2n+1) : One chromosome is added to the diploid set, so that one chromosome occurs in triplicate.
  (d) Tetrasomy (2n+2) : Two chromosomes are added to the diploid set, so that a chromosome is found in quadrapulate.

(b) Polyploidy (Euploidy)

- It is an increase **in** the **number of chromosomes** sets beyond the diploid X condition (2n).
- This is often seen in plants.
- Based on the number of chromosome sets, the polyploid are of the following types : triploids (3n), tetraploids (4n), pentaploids (5n), hexaploids (6n), etc.

(a) Autopolyploidy : It is an increase in number of the same genome. e.g., AAA (autotriploid), AAAA (autotetraploid), etc.

(b) Allopolyploidy : It is the increase in the number of sets of chromosome due to the coming together of diploid genomes of two or more than two individuals of different species. e.g., AABB, AABBDD. Bread wheat is allohexaploid (AABBDD). Triticale is the man-made cereal formed by hybridization between durum, wheat and rye. It is allohexaploid.

- **Autoallopolyploidy :** It is a kind of polyploidy where the genomes of two species come together in which one has double set of chromosomes. e.g., *Helianthus tuberosus* which is autoallohexaploid.
- **Chromosomal aberrations :** These are the changes in morphology and structure of chromosome resulting in the change in number and sequence of genes on them without any change in ploidy. They are of the following types :
  1. **Deletion :** It is the loss of a terminal segment of a chromosome or from within the chromosome (interstitial segment) followed by a reunion of its remaining parts.
  2. **Inversion :** It is a change in a chromosome architecture due to breaking up, rotation through 180° of a segment and its reunion so that sequence of genes is reversed in the inverted region.
  3. **Duplication :** It is a change in chromosome structure in which a part of a chromosome breaks up and unites with another homologous chromosome. This process repeats the chromosome segments because the same block of genes is present more than once in a haploid component.
  4. **Translocation :** It is a change in chromosome architecture that is due to breaking up of segment of chromosome and its union with another non-homologous chromosome. It may also be due to mutual exchange of chromosomal segments between non-homologous chromosomes.

Examples for Chromosomal Disorders

(a) Down's Syndrome (Mongolism) :

- It is the presence of an additional copy of chromosome number 21 (trisomy of 21).
- Genetic constitution : 45 A + XX or 45 A + XY (i.e., 47 chromosomes).
- **Features :**
  (a) They are short-statured with small round head.
  (b) Broad flat face.
  (c) Furrowed big tongue and partially open mouth.
  (d) Many "loops" on fingertips.
  (e) Palm is broad with characteristic palm crease.
  (f) Retarded physical, psychomotor & mental development.
  (g) Congenital heart disease.

Key Fact

Punnett, Batteson and other workers found Mendel's work as an universal application, including animals also.

(b) Klinefelter's Syndrome :

- It is the presence of an additional copy of X-chromosome in male.
- Genetic constitution: 44 A + XXY (i.e., 47 chromosomes).
- **Features :**
  (a) Overall masculine development however the feminine development is also expressed. e.g., development of breast (Gynaecomastia).
  (b) Sterile.
  (c) Mentally retarded.

(c) Turner's Syndrome :

- This is due to the absence of one of the X chromosomes in female.
- Genetic constitution: 44 A + XO (i.e., 45 chromosomes).
- **Features :**
  (a) Sterile, Ovaries are rudimentary.
  (b) Lack of other secondary sexual characters.
  (c) Dwarf.
  (d) Mentally retarded.

Mnemonics

Concept: Chromosomal Disorders

Mnemonics: Dying to **k**now

Interpretations: Down's syndrome Turner's syndrome**,** Klinefelter's syndrome