Chapter 5
Principles of Inheritance and Variation

Genetics: Genetics is a branch of biology which deals with principles of inheritance and its practices.
Heredity: It is transmission of traits from one generation to another through the molecular mechanism.
Variation: Variation is the degree by which progeny differ from their parents.
Allele: Various or slightly different forms of a gene, having same position on chromosomes.

Phenotype: The observable or external characteristics of an organism.

Genotype: The genetic constitution of an organism.

Monohybrid cross : A cross between two individuals of species, considering the inheritance of single pair of contrasting character *e.g.,* a cross between pure tall (TT) and Dwarf (tt).

Dihybrid cross: A cross between two individuals of a species, considering the inheritance of two pairs of contrasting traits/characters *e.g.,* a cross between Round & Yellow seeds (RRYY) and wrinkled & green seeds (rryy).

Incomplete dominance: When one of the two alleles of a gene is incompletely dominant over the other allele.

Co-dominance: When two alleles of a gene are equally dominant and express themselves even when they are together.

Multiple allelism : When a gene exists in more than two allelic forms *e.g.,* gene for human blood group exist in three allelic forms, I^A, I^B and i.

Aneuploidy : The phenomenon of gain or loss of one or more chromosome(s), that results due to failure of separation of homologous pair of chromosomes during meiosis.

Trisomy : The condition in which a particular chromosome is present in three copies in a diploid cell/nucleus.

Male heterogamety : When male produces two different types of gametes/sperms *e.g.,* In human beings X and Y.

Female Heterogamety : When female produces two different types of gametes/ova *e.g.,* female bird produces Z and W gametes.

Mutation : The sudden heritable change in the base sequence of DNA, or structure of chromosome or a change in the number of chromosomes.

Pedigree Analysis : The analysis of the distribution and movement of trait in a series of generations of a family.

Law of Dominance: i) Characters are controlled by discrete units called factors. Ii) Factors occur in pairs. iii) In a dissimilar pair of other (recessive).

Law of Segregation: The members of allelic pair that remained together in the parent Segregate/separate during gamete formation and only one of the factors enters a gamete.

Law of Independent Assortment: In the inheritance of two pairs of contrasting characters, the factors of each pair of characters segregate independently of the factors of the other pair of characters.

Test Cross: When offspring or individual with dominant phenotype, whose genotype is not known, is crossed with an individual who is homozygous recessive for the trait.

Aneuploidy: Failure of segregation of chromatids during cell division results in the gain or loss of a chromosome(s) called aneuploidy.

Polyploidy: Failure of cytokinesis after telophase stage of cell division results in an increase in a whole set of chromosomes in an organism and this phenomenon is known as polyploidy.

Pleiotropism: Pleiotropism is defined as a phenomenon when single gene may produce more than one effect (the multiple effect of a gene) or control several phenotypes depending on its position.

Carrier: A person with a 'defective recessive gene' and a 'dominant normal gene' on homologous pair of chromosomes and therefore, not affected by **the disorder but transmit the defective gene to the next progeny through gametes.**

Genome: All the genetic material in the chromosomes of a particular organism; size generally given as its total number of base pairs.

Germ Cell- An egg or sperm cell. A gamete. In humans, a germ cell contains 23 chromosomes.

Haploid= A single set of chromosomes (half the full set of genetic material), present in the egg and sperm cells of animals and in the egg and pollen cells of plants. Human beings have 23 chromosomes in their reproductive cells.

Hemophilia = sex-linked recessive. Males get it most often.

Homologous chromosomes: A pair of chromosomes containing the same linear gene sequences, each derived from one parent

Karyotype: Photomicrograph of an individual's chromosomes arranged in a standard format showing the number, size, and shape of each chromosome type.

Linkage: Proximity of two or more genes on a chromosome. The closer together the genes, the lower the probability that they will be separated during meiosis and hence the greater the probability that they will be inherited together.

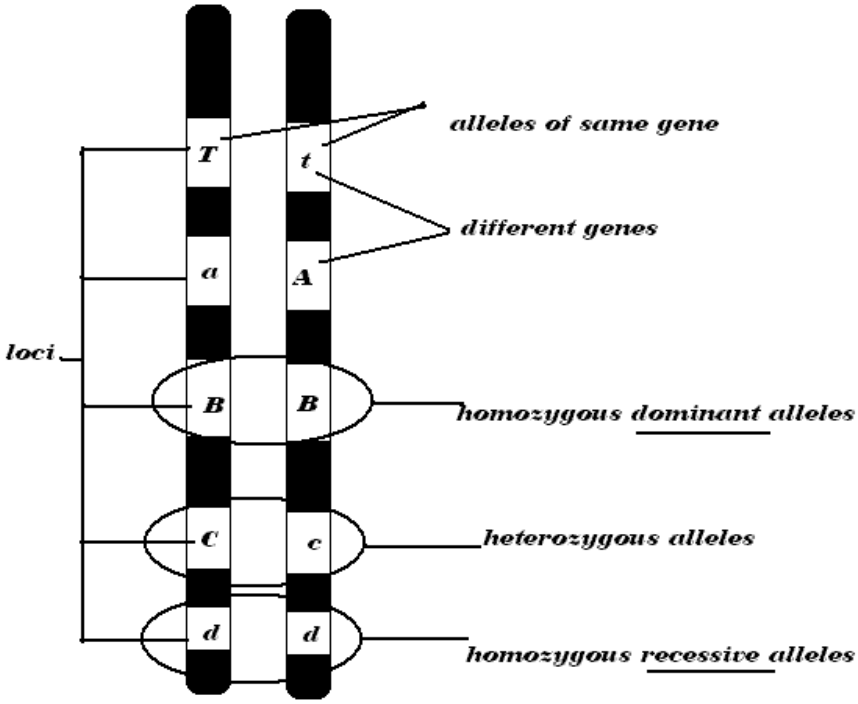
Linkage map: relative positions of genetic loci on a chromosome, determined on the basis of how often the loci are inherited together.

Locus (pl. loci): The position on a chromosome of a gene or other chromosome marker; also, the

DNA at that position. The use of locus is sometimes restricted to mean regions of DNA that are expressed.

Non-Disjunction: When homologous chromosomes fail to segregate properly during meiosis. Down syndrome, Turner syndrome and Klinefelter syndrome result from non-disjunction.

Relationship between genes and chromosome of diploid organism and the terms used to describe them



Understand the following terms with the help of the diagram shown above.

Terms	Meaning	Example
Locus	Address/ location of a gene in a chromosome	T,A.b,d etc
Allele	Allelomorphs= alternative form of a gene	T and t OR A and a etc
Homozygous	Both alleles of a gene at a locus similar	AA or aa
Heterozygous	Both alleles of a gene at a locus dissimilar	Aa or Tt etc
Homozygous Dominant	Both alleles of a gene at a locus similar & dominant	AA
Homozygous recessive	Both alleles of a gene at a locus similar & recessive	aa

Exceptions to Mendel's laws of inheritance

I. Incomplete Dominance

When the dominant allele does not completely the phenotypic expression of the recessive allele in a heterozygote, then a blending of both dominant and recessive traits takes place in F1 and F2 hybrids. This phenomenon is called incomplete dominance.

e.g.i) In snapdragon, broad leaf is incomplete dominant over narrow leaf.

ii) In snapdragon,(or Antirrhinum sp./dog flower) red flower is incompletely dominant over white flower

iii) In Mirabilis, red flower is incompletely dominant, over white flower.

II. Co-dominance (Multiple allelism)

When both dominant and recessive allele lack dominant recessive relationship, then both are expressed side by side. This phenomenon is known as co-dominance.

e.g. ABO blood groups in human beings are controlled by the gene *I.* The gene (I) has three alleles I^A, I^B and i. Since there are three different alleles, there are six different combinations of these three alleles that are possible a total of six different genotypes of the human ABO blood types.

III. Polygenic Inheritance

At least in some instances the same character can be determined by more than one gene, each with the same but cumulative phenotypic effect Quantitative characters like ***plant height, yield of crops (size, shape and number of seeds and fruits per plant), intelligence in human beings and milk yield in animals*** have been found to be determined by many genes and their effects have been found to be cumulative. This phenomenon is known as polygenic inheritance. This is also considered as " Quantitative inheritance" or "multiple factor inheritance".
Other examples that can be studied are the kernel colour in wheat and inheritance of cob length in maize.

IV. Pleiotropism

Pleiotropism is defined as a phenomenon when single gene may produce more than one effect (the multiple effect of a gene) or control several phenotypes depending on its position.

Eg.In drosophila white eye mutation leads to depigmentation in many other parts of the body, giving a pleitropic effect.
In transgenic organisms, the introduced gene can produce different effects depending on where the gene has introgressed.

Some useful information

Cross	Result of F2 generation	
	Phenotypic ratio	Genotypic ratio
Monohybrid cross Tt X Tt	3:1	1:2:1
Dihybrid cross YyRr X YyRr	9:3:3:1	1:2:1:2:4:2:1:2:1

Incomplete dominance Rr X Rr	1:2:1	1:2:1

Co-dominance / multiple allelisim

Blood group	Possible genotype
A	I^AI^A OR I^Ai
B	I^BI^B OR I^Bi
AB	I^AI^B
O	ii

Crosses of blood group (CO-DOMINANCE)

Blood group	Possible genotype	Possible phenotype
A X A	I^AI^A X I^AI^A	A
	I^AI^A X I^Ai	A
	I^Ai X I^Ai	A ; O
B X B	I^BI^B X I^BI^B	B
	I^BI^B X I^Bi	B
	I^Bi X I^Bi	B; O
AB X AB	I^AI^B X I^AI^B	AB: A; B
AxB	I^AI^A X I^BI^B	AB
	I^Ai X I^BI^B	AB,B
	I^AI^A X I^Bi	A,AB
	I^Ai X I^Ai	A,B,AB,O
O X O	ii X ii	O

CHROMOSOMAL THEORY OF INHERITANCE

Sutton and Boveri argued that the pairing and separation of a pair of chromosomes would lead to the segregation of a pair of factors they carried. Sutton united the knowledge of chromosomal segregation with Mendelian principles and called it the "***chromosomal theory of inheritance".***

Linkage and Recombination

The Mendel's laws were extended in the form of 'Chromosomal Theory of Inheritance'. Later, it was found that Mendel's law of independent assortment does not hold true for the genes that were located on the same chromosomes. These genes were called as ***'linked genes'.*** Closely located genes assorted together, and distantly located genes, due to ***recombination,*** assorted independently. Linkage maps, therefore, corresponded to arrangement of genes on a chromosome.

- ***T.H. Morgan : Father of experimental genetics***
- ***Alfred Sturtevant: Used the " frequency of recombination" between gene pairs on the same chromosome as a "measure of distance" between genes and "mapped' their position on the chromosome.***

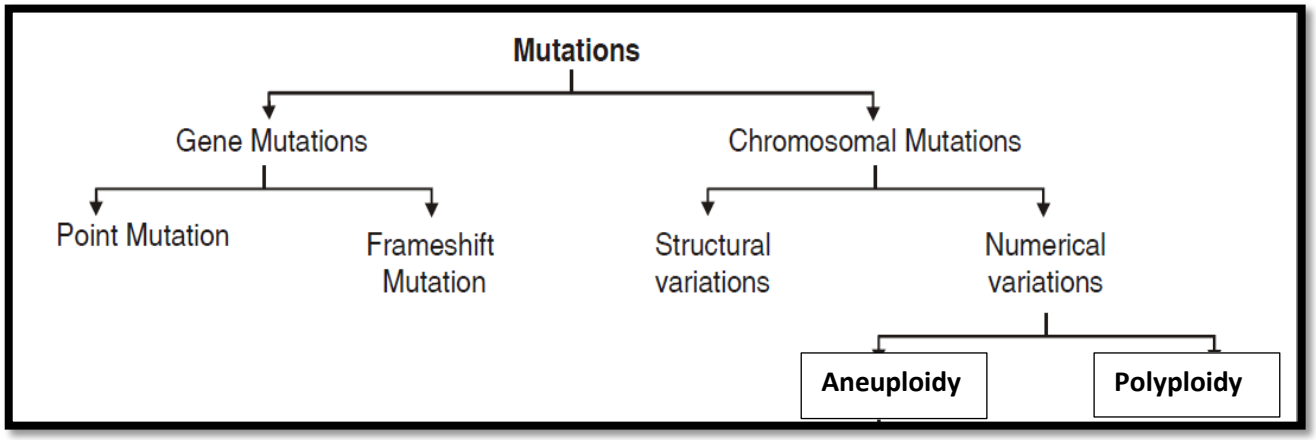
Sex Determination

- The term sex refers to sexual phenotype. Most organisms have only two sexual phenotypes: male and females.
- Sex determination: The mechanism by which sex is established is termed as sex determination.
- **Henking**(1891) was the first to identify the sex chromosome as **'X body'**, but he could not explain its significance.
- Male heterogamety and female heterogamety (refer key point definitions mentioned above) are commonly observed patterns that decide the sex of an organism.

Sex determination and sex chromosome

Organism	Male	Female
Human beings, *Drosophila*	XY	XX
Many Birds	ZZ	ZW
Insects(Grasshopper)	XO	XX

Mutation



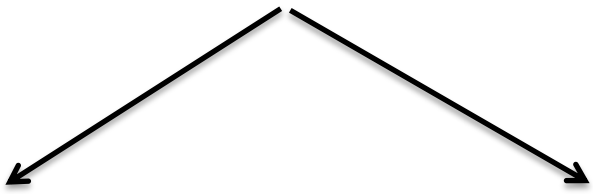
Pedigree Analysis

Pedigree Analysis : The analysis of the distribution and movement of trait in a series of generations of a family.

Note: Refer NCERT Text Book Page No. 88 for the **symbols used in the human pedigree analysis.**

Autosomal Dominant	Autosomal Recessive
1. Traits are controlled by dominant genes 2. Both males and females are equally affected **3. Traits do not skip generations** 4. e.g. Myotonic dystrophy	1. Traits controlled by recessive genes and appear only when homozygous 2. Both male and female equally affected **3. Traits may skip generations** 4. 3:1ratio between normal and affected. 5. Appearance of affected children from normal parents (heterozygous) 6. All children of affected parents are also affected. 7. e.g.- Sickle-cell anaemia

GENETIC DISORDERS



MENDELIAN DISORDERS

(Autosome- linked/Sex-linked)

Haemophilia, Cystic fibrosis, Sickle-cell anaemia, colour-blindness,

CHROMOSOMAL DISORDERS

Down's Syndrome(Trisomy of 21^{st})

Klinefelter's Syndrome(XXY in male)