Heredity and Variation:

An organism produces an organism of similar kind. All the offspring are similar with their parents in many respects. This property of an individual to resemble with their parents is called heredity. In other words, heredity is the passing of traits to offspring. This is the process by which an offspring acquires or becomes predisposed to the characteristics of its parent. It may be defined as transmission of characters from parents to offspring.

Although offspring resemble with their parents in many respects, there are not exactly similar in sexual reproduction. The difference between parents and offspring or among the offspring of the same parents is called variation.

Types of Variation:

There are found various types of variations. They are as follows:

1. **Variation based on the nature of cell it affects:**
   On the basis of nature of cell it affects, variations are divided into two types.
   a. **Somatic variation:** It occurs in vegetative cell of the organism. Such variations are acquired by organism during its own life time and lost with the death. It arises due to the environmental effect and use and disuse of organs.
   b. **Germinal variation:** It occurs due to the change in germ cells. It is inheritable i.e. transfer the character from parents to offspring. It arises due to crossing over, mutation, radiation, recombination of genes etc.
2. **Variation based on degree of difference:**
   On the basis of degree of difference, variation is of two types.
   a. **Continuous variation:** It is small and indistinct variation found on organism during a long course of time. It is gradual, non-distinct. They are non-inheritable. eg: change in weight, size, colour etc.
   b. **Discontinuous variation:** It is large and distinct characters of offspring which are different from parents. It occurs due to the mutation which seen suddenly and is stable. It is inheritable. eg: hairless variety of dogs and cats, polydactyly etc.

Difference between clone and offspring

Clone	Offspring
1. It is derived from single parent.	1. It is formed from two parents.
2. It is produced from asexual reproduction.	2. It is produced from sexual reproduction.
3. It is a production of mitosis only.	3. Meiosis occurs to the formation of gametes.
4. Recombination of gene does not take place.	4. Recombination of gene regularly occurs.

5. It does not involve in the formation of gametes. 6. It is exactly like the parent.	5. It involves the formation and fusion of gametes. 6. It is different from the parents.

Some terms used in Genetics:

Gene: A small segment of DNA which determines only one character of an organism.

Locus: The specific position occupied by a gene on a specific chromosome is called locus.

Alleles: Two or more alternative forms of a gene are called alleles.

Genotype: Genetic makeup of an organism or list of genes present in an organism is called genotype.

Phenotype: The externally appeared and physiological character of an organism is called phenotype.

Homozygous: An organism having two similar alleles of a gene is called homozygous.

Heterozygous: An organism having two different alleles of a gene is called heterozygous.

Monohybrid: An individual which is hybrid for only one gene.

Dihybrid: An individual which is hybrid for two genes.

Clone: A group of genetically identical cells or organisms derived from a single parent by asexual reproduction.

Mutation: A sudden heritable change in the genetic material of an organism is called mutation.

Parents: The organisms which are used for a cross.

Offspring: The individuals resulting from a cross.

Gene pool: The total variety of genes and alleles present in sexually reproducing population is called gene pool.

Gene frequency: The number of organisms in a population carrying a particular gene or alleles is called gene frequency.

Mendel's Experiment:

Mendel used pea plant for hybridization experiments because of following reasons

1. Pea plant contains many varieties with well defined characters. Mendel selects seven different unit characters for his experiments.
2. Flower is bisexual where stamen and pistil enclosed within corolla.
3. They have very short life cycle.
4. Large number of seeds can be formed.
5. Hybrid plant is again fertile.
6. They could be easily self pollinated or cross pollinated.

7. They are easily grown and does not require extra cure.
8. They are medium sized.

Monohybrid Cross: The cross between two homozygous or pure parents differing in only one pair of contrasting characters.
Chart
Dihybrid Cross: The cross between two homozygous or pure parents differing in two pairs of contrasting characters.
Chart

Mendel's Laws of Inheritance:
Mendel formulated three laws on the basis of crosses in pea plant.

1. **Law of Dominance:** When two homozygous parents differing in only one pair of contrasting characters are crossed, only one character is expressed in F_1 generation. The character which is expressed is called dominant character while the other is not expressed is called recessive character.
2. **Law of Segregation:** The heterozygous individual of F_1 generation contains two contrasting allelic factors of a pair of contrasting characters. They remain together for a long time but do not mix with each other. They separate at the time of gamete formation so that each gamete receives only one allelic factor. Due to such property of gametes, this law is also known as law of purity of gametes.
3. **Law of Independent Assortment:** The inheritance of a pair of contrasting characters is independent of another pair of contrasting characters.

Punnett Square: It is a check board used to study all possible results of various crosses. It is first developed by R.C. Punnett (1906).

Nucleic Acid:
Nucleic acids are non-protein, nitrogenous substances which are macromolecule composed of chains of nucleotides. These molecules carry genetic information within cells. The most common nucleic acids are deoxyribonucleic acid (DNA) and ribonucleic acid (RNA). Nucleic acids are universal in living things. The nucleic acids DNA and RNA are made up of pentose sugar, nitrogenous bases and phosphoric acid.

1. **Pentose sugar:** It is the type of sugar that contains 5 carbon atoms. It is of two types.
   a. **Ribose sugar:** It is found in RNA. It is water soluble pentose sugar that is an important component of nucleic acids, nucleotides, the vitamin riboflavin and various co-enzymes. It has the chemical formula $C_5H_{10}O_5$.

   b. **Deoxyribose sugar:** The deoxyribose sugar present in DNA. It is the pentose sugar that lacks one oxygen atom from second carbon of ribose sugar. It has the chemical formula $C_5H_{10}O_4$.
2. **Nitrogenous bases:** Nitrogenous bases are an organic compound that owes its property as a base to the lone pair of electrons of a nitrogen atom. They are non-polar and due to their aromaticity, planar. They are typically classified as the derivatives of two parent compounds pyrimidine and purine.
   a. **Pyrimidine:** It is a heterocyclic aromatic organic compound similar to benzene and pyridine, containing two nitrogen atoms at positions 1 and 3 of the six member ring.
      It is of three types. They are cytosine, thymine and uracil.
   b. **Purine:** It is a heterocyclic aromatic organic compound consisting of a pyrimidine ring fused to an imidazole ring. Purines are the most widely distributed kind of nitrogen containing heterocyclic in nature.
      It is of two types. They are adenine and guanine.
3. **Phosphoric acid:** It contains phosphate group.

Nucleoside: It is formed by the combination of purine or pyrimidine base linked to pentose sugar.
Nucleotide: It is formed by the combination of nucleoside with phosphoric acid.

DNA (Deoxyribose Nucleic Acid):
It is a nucleic acid that contains the genetic instructions used in the development and functioning of all known living organisms. The main role of DNA molecules is storage of information. The DNA segments that carry this genetic information are called genes.
DNA consists of two long polymers of simple units called nucleotides with backbones made up of sugars and phosphate groups joined by ester bonds. These two strands run in opposite directions to each other and are therefore antiparallel.
Within cells, DNA is organized into long structures called chromosomes, nucleolus, mitochondria and chloroplast.

Structure of DNA:
Watson and Crick proposed the DNA model by using all the information that was available at that time. They used the data obtained from experiments carried out on DNA.
The important features of this model are:
1. The DNA molecule is a double helix with single polynucleotides running in opposite directions.
2. The double helix is right handed.

3. The double helix has two different grooves.
4. The nitrogenous bases are stacked towards the inside of the helix.
5. Bases of the two polynucleotides interact by hydrogen bonding. An adenine always adjacent to thymine; guanine is always adjacent to cytosine.
6. Ten base pairs occur per turn of the helix. The height or pitch of the helix is $34A^o$. The space between two base pairs to opposite strand is $8.4A^o$ and has an angle of 36^o.
7. The diameter of the helix is $20A^o$.

Function of DNA:

1. It acts as a carrier of genetic information from generation to generation.
2. It synthesizes ribonucleic acid (RNA).
3. It acts a prime molecule during protein synthesis.
4. It controls off the biological activities of cell.
5. DNA has autocatalytic function which directs the synthesis of its own copy.

RNA (Ribose Nucleic Acid):

It is a nucleic acid that contains a polymer of ribonucleotides of adenine, uracil which are joined together by phosphodiester bond. It is found in nucleolus, ribosomes, mitochondria, chloroplast and cytoplasm.

Structure:

1. A molecule of RNA has only one polynucleotide chain.
2. The polynucleotide chain is made up of ribonucleotides attached to each other by 3'-5' phosphodiester bond.
3. The backbone of the strand is made up of pentose sugar and phosphate while the nitrogen bases form backbone towards the axis.
4. Pentose sugar is ribose.
5. An RNA molecule may contain self complementary sequences that allow parts of the RNA to fold and pair with itself to form double helices.

Differences between DNA and RNA

DNA	RNA
1. It is double stranded.	1. It is single stranded.
2. It is spirally coiled to form double helix structure.	2. It is linear or partially coiled.
3. It contains deoxyribose sugar as pentose sugar.	3. The pentose sugar is ribose sugar.
4. Nitrogen bases are adenine, guanine, cytosine and thymine.	4. Nitrogen bases are adenine, guanine, cytosine and uracil.

5. Complementary base pairing present.	5. Complementary base pairing may or may not be present.
6. The ratio of purine and pyrimidine is 1.	6. The ratio of purine and pyrimidine is not equal to 1.
7. It acts as genetic material of all living organisms.	7. It helps in protein synthesis as well as generic material in RNA virus.

Genetic Diseases:

Human beings suffer from many genetic diseases or disorders. They arise in human by different ways. They are chromosomal abnormalities, gene mutation and gene incompatibility.

1. **By chromosomal abnormalities:** There are two types of abnormalities found in autosomes and sex chromosome. They are named as autosomal abnormalities and sex chromosome abnormalities.

   A. **Autosomal abnormalities:** The abnormalities found in autosomes are known as autosomal abnormalities. They are of following types.

      a. **Down's Syndrome:** It is also known as Mongolian Idiocy or Mongolism or 21 trisomy. It was reported by Langdon Down in 1866. It is caused due to presence of an extra chromosome number 21. Thus the offspring has 47 chromosomes (45+XY) in male and (45+XX) in female. One in every 600 children may victimized by this disease.

         Symptoms:

         i. Affected children have broad forehead, short neck and flat heads.
         ii. They have permanently opened mouth and extended tongue.
         iii. Their brain is formed with little intelligence.
         iv. They have slanting eyes with folded eyelid.
         v. Heart and other organs may be defective.

      b. **Patau's Syndrome:** It is also known as13 trisomy. It was reported by Patau in 1916. It is caused due to presence of an extra chromosome number 13. One in every 5000 children may victimized by this disease. The average life span of affected person is about 4 months.

         Symptoms:

         i. The person has small head and abnormalities of the face, eyes, cleft, lip and palate.
         ii. Prominence of the posterior part of the head, deafness, mental deficiency, malformation of external and internal organs etc.

      c. **Edward's Syndrome:** It is also known as18 trisomy. It was reported by Edward in 1960. It is caused due to presence of an extra chromosome

number 18. It occurs in about one in 3500 live births. It is more common in female than in male.

Symptoms:

i. The victim keep the fingers tightly clenched (closed) against the palm of the hand.
ii. They have small jaws, deformed (misshapen) ears, small sternum and pelvis.
iii. They are mentally retarded.

d. Cri-du-chat Syndrome: It is also known as cat cry because the affected child cries like mewing of a cat. It was reported by Lejeune in 1963. It is caused due to the deletion of one arm of the 5^{th} chromosome. It is very rare.

Symptoms:

i. Affected child has widely spaced eyes, small head and receding chin (slope backwards).
ii. They may suffer from congenital heart disease as well as various physical and mental retardations.

B. Sex chromosome abnormalities: The abnormalities found in sex chromosome are known as sex chromosome abnormalities. They are of following types.

a. Klinefelter's Syndrome: It is caused due to the presence of an extra chromosome in male. Such males have 47 chromosomes with extra X chromosome i.e. 44+XXY. It occurs in about one in 2000 live births.

Symptoms:

i. The affected person is a sterile male with small testes, unusual long legs and sparse body hairs.
ii. Such person possesses some female characters like enlarged breast.
iii. The victim has abnormalities in skeleton and low mental ability.

b. Turner's Syndrome: It is caused due to absence of one X chromosome in female. Such females have 45 chromosomes i.e. 44+X0. This occurs in about one in 5000 births.

Symptoms:

i. They are sterile females with poorly developed ovaries and underdeveloped breasts.
ii. They have webbed neck and broad chest.
iii. They possess many male characters like heavy neck muscles and narrow hips.
iv. They are short stature (height) without puberty and menstrual cycle.

c. **Super males:** It is also known as Jacob's syndrome. It is caused due to the presence of extra chromosomes in male. Such males have 47 chromosomes with extra Y chromosome i.e. 44+XYY. It occurs in about one in 1000 boys.

Symptoms:

i. They have abnormal height, mental retardation and criminal bent of mind.
ii. They have over production of male sex hormone.
iii. They are more aggressive than normal males.

d. **Super females:** It is also known as trisomy X or triple X syndrome. It is caused due to presence of extra chromosomes in female. Such female have 47 chromosome with extra X chromosome i.e. 44+XXX. It occurs in about one in 1000 live births.

Symptoms:

i. They are tall stature, small head.
ii. They have learning disabilities or weak muscle tone.
iii. They tend to show accelerated growth until puberty.

2. **By gene mutation in chromosome:** There are two types of mutation found in autosomes and sex chromosome. They are known as gene mutation in autosome and gene mutation in sex chromosome.

A. Gene mutation in autosomes: It is again of two different types. They are as follows:

I. Recessively inherited traits: It is due to presence of recessive genes in homologous condition. Some disorders of this type are as follows

a. **Alkaptonuria:** It is also known as black urine disease, black bone disease. It was reported by Garrod in 1908. It is an inborn metabolic disorder. It is caused by the deficiency of an oxidase enzyme (It is required for break sown of homogentisic acid. It accumulates in tissue and excreted in the urine).

Symptoms:

i. The urine becomes black on exposure to air.
ii. The accumulation of homogentisic acid affects the cartilages, capsules of joints, ligaments and tendons.
iii. It also causes arthritis.

b. **Phenylketonuria (PKU):** It occurs due to lack of enzyme phenylalanine hydroxylase. This enzyme is needed to change amino acid phenylalanine to tyrosine. Hence, the phenylalanine accumulates in the tissue and later excreted in urine.

Symptoms:

i. It leads mental retardation in children.
ii. It decreases pigmentation in skin and hair.
iii. They may be unable to walk and talk it not treated in time.

c. Albinism: It is occurred due to absence of enzyme tyrosinase. This enzyme is essential for the synthesis of melanin pigment from dihydroxyphenyl-alanine.

Symptoms:

i. They lack dark pigment, melanin, in the skin, hair and iris.
ii. It causes poor vision, but they show normal life.

d. Tay-sach's disease: It is inborn disease but the children with this disease born normal. This develops later. It leads to severe damage to brain and spinal cord with few months. They become mentally retarded and paralysed within 3—4 years that leads to death.

e. Sickle cell anemia: It is caused by the presence of pleiotropic gene which shows effect RBC causing sickle cell anemia. This is caused by a gene which is lethal in homozygous condition that leads to die.

II. Dominantly inherited traits: It is due to presence of dominant genes. These disorders are

a. Dwarfism
b. Polydactyly
c. Brachydactyly

B. Gene mutation in sex chromosome: They are produced by changes in the genes lying in the sex chromosome. These are called sex linked disorders. The sex linked disorders are followings

a. Haemophilia (Bleeder's disease)
b. Colour blindness
c. Night blindness

3. By gene incompatibility: Some disorders occur in the offspring due to gene product in the blood. It leads to destruction of foetal blood cells commonly known as Rh-factor incompatibility.

a. Blood transfusion
b. Pregnancy

Determination of Sex:

Autosomes: The chromosomes which are identical and control morpho-physiological characters are autosomes. They are similar in both of the sexes.

Sex chromosome: Those chromosome which singly or in pair and responsible to determine sex of individual are called sex chromosome. They are differ in different sexes.

Types of Sex determination:

Sex is the most important hereditary differences found between two individuals of same species. Sex determination is determined at the time of fertilization. It is divided into three types.

1. **Environmental or non-genetic determination of sex:** In some species environment determine the sex.
   In the European pond turtle, incubation temperature above 30°C produces all females whereas below 25°C produces all males. At 28°C, equal number of males and females are produced.
2. **Non-chromosomal genic determination of sex:** In bacteria, there are present non-chromosomal genetic segments known as fertility factor in plasmid determines the sex. If fertility factor is present, it represents male strain otherwise female.
3. **Chromosomal determination of sex:** In most animals, sex is genetically determined. It is based on the formation of two types of gametes in one sex i.e. heterogametogenesis. It is studied under two headings.
   - **A. Male heterogamy:** In certain organisms, male forms two types of gametes i.e. one is X and other lacks it. It is again divided into following headings.
     - **a. XX-XY type:** It is found in mammals and most insects like fruit fly, *Drosophila*.
       Human 44+XX 44+XY
       Fruit fly 6+XX 6+XY
     - **b. XX-XO type:** It is found in some insects like cockroach and some roundworms.
       AA+XX AA+XO
   - **B. Female heterogamy:** In some organisms like fishes, reptiles, birds and some insects like moth, the female produce two types of gametes. It is again divided into following headings.
     - **a. ZW-ZZ type:** It is found in some birds like domestic fowl, butterflies and some fishes.
       AA+ZW AA+ZZ
     - **b. ZO-ZZ type:** It is found in certain moth and butterflies.
       AA+ZO AA+ZZ