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.
   Chart:
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.
   Chart:
3. **Law of Independent Assortment:** The inheritance of a pair of contrasting characters is independent of another pair of contrasting characters.
   Chart:

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).

Genetic Code:

The genetic code is the set of rules by which information encoded in genetic material (DNA or mRNA sequences) is translated into proteins (amino acid sequences) by living cells. The code defines from codons and amino acids. A triplet codon in a nucleic acid sequence specifies a single amino acid. Because the vast majority of genes are encoded with exactly the same code, this particular code is often referred to as the canonical or standard genetic code, or simply the genetic code, though in fact there are many variant codes.

Genetic Code Table:

First base	Second base				Third base
	U	C	A	G	
U	UUU, UUC } Phe UUA, UUG } Leu	UCU, UCC, UCA, UCG } Ser	UAU, UAC } Tyr UAA, UAG } Stop	UGU, UGC } Cys UGA } Stop UGG } Trp	U C A G
C	CUU, CUC, CUA, CUG } Leu	CCU, CCC, CCA, CCG } Pro	CAU, CAC } His CAA, CAG } Gln	CGU, CGC, CGA, CGG } Arg	U C A G
A	AUU, AUC, AUA } Ile AUG } Met	ACU, ACC, ACA, ACG } Thr	AAU, AAC } Asn AAA, AAG } Lys	AGU, AGC } Ser AGA, AGG } Arg	U C A G
G	GUU, GUC, GUA, GUG } Val	GCU, GCC, GCA, GCG } Ala	GAU, GAC } Asp GAA, GAG } Glu	GGU, GGC, GGA, GGG } Gly	U C A G

Properties of Genetic Code:

1. Code is triplet
2. Code is non-overlapping
3. Code is commaless
4. Code is universal
5. Code is degenerate
6. Unambiguity of code
7. Initiation codon
8. Nonsense codon
9. Colinearity

Mutation: Mutations are changes in a chromosomal sequence. Mutations are caused by radiation, viruses and mutagenic chemicals, as well as errors that occur during meiosis or DNA replication. They can also be induced by the organism itself, by cellular processes such as hypermutation.
Mutation can result in several different types of change in DNA sequences; these can have no effect, alter the product of a gene, or prevent the gene from functioning properly or completely.

Mutation is of two types.

1. **Gene mutation:** A gene mutation is a permanent change in the DNA sequence that makes up a gene. Mutations range in size from a single DNA building block (DNA base) to a large segment of a chromosome. It is also known as point mutation or micro-mutation.
   Gene mutations occur in two ways: they can be inherited from a parent or acquired during a person's lifetime. Mutations that are passed from parent to child are called hereditary mutations or germline mutations. This type of mutation is present throughout a person's life in virtually every cell in the body.
   Different types of gene mutations are given below
   a. **Frame-shift mutation:** A mutation in a DNA chain that occurs when the number of nucleotides inserted or deleted is not a multiple of three, so that every codon beyond the point of insertion or deletion is read incorrectly during translation.
   b. **Substitution mutation:** Mutation which arises due to replacement of one base by another base is called substitution mutation. It changes only one or few codon. It does not change the reading frame of genetic code. It is again two types.
      i. **Transition Mutation:** Transition Mutation is a point mutation that changes a purine nucleotide to another purine (A ↔ G) or a pyrimidine nucleotide to another pyrimidine (C ↔ T). Approximately two out of three single nucleotide polymorphisms are transitions.
      ii. **Transversion:** Transversion refers to the substitution of a purine for a pyrimidine or vice versa. Transversions can be caused by ionizing radiation and alkylating agents.
2. **Chromosomal Mutation:** A mutation involving a long segment of DNA. These mutations can involve deletions, insertions, or inversions of sections of DNA. In some cases, deleted sections may attach to other chromosomes, disrupting both the chromosomes that lose the DNA and the one that gains it.
   It is also divided into two types.
   a. **Chromosomal Aberrations:** Chromosomal aberrations are abnormalities in the structure or number of chromosomes and are often responsible for genetic

disorders. For more than a century, scientists have been fascinated by the study of human chromosomes.

It is of following types.

i. **Deficiency:** A deficiency is the loss of a segment of a chromosome. The amount of deficiated material may be any length from a single base to a large piece of the chromosome. Only small deletions are tolerated. Larger deletions and the deficiated of an entire chromosome always result in nonviable embryos.

ii. **Duplications:** Duplications also result from the reuniting of broken pieces of homologous chromosomes. In some cases the chromosome pieces rejoin in such a way that there is a doubling, or redundancy, of a portion of the chromosome. This changes the number of genes present and may result in a problem with health, development, or growth.

iii. **Inversions:** An inversion is the rotation of a broken chromosome segment in such a way that it rejoins the chromosome in a reversed state, or is flipped, end to end.

iv. **Translocation:** If the broken end of a chromosome joins to another non homologous chromosome, such aberration is called translocation. An exchange of parts between two non homologouos chromosomes is known as reciprocal translocation. If a part of chromosome is translocated to another non homologouos chromosome, it is called non-reciprocal trasnloction.

b. **Ploidy:** Mutation that takes place due to change in number of chromosome is called ploidy. It is of following types.

i. **Aneuploidy:** Aneuploidy is an abnormal number of chromosomes in the zygotes, and is a type of chromosome abnormality. An extra or missing chromosome is a common cause of genetic disorders (birth defects). Some cancer cells also have abnormal numbers of chromosomes. Aneuploidy occurs during cell division when the chromosomes do not separate properly between the two cells. It is following types.

- **Monosomics:** The diploid organisms which lack in one chromosome of a single homologous pair of chromosome are called monosomics with the genomic formula 2n -1. A monosomic produces two kinds of gametes, (n) and (n - l).
- **Nullisomics:** The diploid organisms which have lost a pair of homologous chromosomes are called nullisomics with the genomic formula, 2n-2.
- **Trisomics:** The diploid organisms which have one extra chromosome are called trisomies. They have the chromosomal

formula 2n+1. In a trisomic, one of the pairs of chromosomes has an extra member, therefore, forms a trivalent structure during meiosis.

- **Tetrasomics:** When one chromosome of an otherwise diploid organism is present in quadruplicate, the tetrasomy is resulted. The tetrasomics have the chromosomal formula 2n+2. During meiosis a quadrivalent is formed by extra chromosomes and segregation of chromosomes occurs like autotetraploids.

ii. **Euploidy:** It is the condition having a chromosome number that is an exact multiple of the haploid number for the species.

- **Monoploidy:** A multiple of the basic number of chromosomes in a cell. During meiosis the cell produces sex cells (gametes), each containing half the normal number of chromosomes, a condition called haploidy.
- **Polyploidy:** It is the condition where more than two sets of chromosomes. Polyploidy may be
  - Triploid
  - Tetraploid
  - Pentaploid
  - Hexaploid

Causes of Mutation: Mutations occurs various agents. Such agents are called mutagens. They are as follows:

a) Chemical Mutagens: There are various chemicals which cause mutation. Such as

i) Tautomerism: When a molecule can exist more than one chemical forms, it is called taotomeric and the process is called tautomerism. Tautomerism changes the pairing ability of purine with pyrimidine of vice versa.

ii) Deamination: The removal of NH_2 group from a compound is called deamination. It replaces NH_2 group by OH group in nitrogen bases and changes structure of nitrogen bases. It is cause due to the chemical nitrous oxide (HNO_2).

iii) Methylation: The process of addition of methyl group to nitrogen bases. It is cause due to the chemical $(CH_2Cl_2)_2$.

b) Physical Mutagen: Different kinds of rays such as X-rays, alpha rays, gamma rays, beta rays etc produce change in chemical structure of gene. High energy of radiations usually breaks the DNA. Very low or high temperature also causes mutation. Similarly, radium UV rays are also mutagens.

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. Eg: Down's syndrome, Patau's syndrome, Edward's syndrome, Cry-du-chat syndrome, Wolf-Hirschhorn syndrome, Jacobsen syndrome, etc.
   - **B. Sex chromosome abnormalities:** The abnormalities found in sex chromosome are known as sex chromosome abnormalities. Eg: Klinefelter's syndrome, Turner's syndrome, Super males, Super females, etc.
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. Eg: Alkaptonuria, Phenylketonuria, Albinism, Tay-sach's disease, Sickle cell anemia, etc.
     - **II. Dominantly inherited traits:** It is due to presence of dominant genes. Eg: Dwarfism, Polydactyly, Brachydactyly, etc.
   - **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. Eg: Haemophilia (Bleeder's disease), Colour blindness, Night blindness, etc.
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. Eg: Blood transfusion, Pregnancy, etc.
4. **Multi factorial genetic disorder:** These are caused by multiple genes come together along with the effects of environmental factors. Eg: Diabetes, Asthma, High blood pressure, Alzheimer's disease, Obesity, Epilepsy, Heart diseases, Hypothyroidism, Autism, Club foot, Cancer (breast, ovaries, bowel, prostate, skin), Birth defects (neural tube defects, cleft palate), even Dandruff, etc.

Some genetic disorders:

1. **Down's Syndrome:** It is also known as Mangolian 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. They are short stature.
ii. Affected children have broad forehead, short neck and flat heads.
iii. They have slanting eyes with folded eyelid.
iv. They have flat nose bridge, small ears, hands and feet.
v. They have permanently opened mouth and extended tongue.
vi. Their brain is formed with little intelligence.
vii. Small finger that point towards the thumb.
viii. Heart and other organs may be defective.

2. **Edward's Syndrome:** It is also known as 18 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. They are small physical sized (head, mouth and jaw).
ii. They have underdeveloped bodies.
iii. The victim keep the fingers tightly clenched (closed) against the palm of the hand.
iv. They have small jaws, deformed (misshapen) ears, small sternum and pelvis.
v. They have minimum response to sound.
vi. They may suffer from congenital heart disease and kidney disease.
vii. They are mentally retarded which are typically severe.

3. **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, short trunk and sparse body hairs.
ii. They have radioulnar synostosis and flat feet.
iii. Such person possesses some female characters like enlarged breast.
iv. They have weaker bones and higher risk of fractures.
v. The victim has abnormalities in skeleton and low mental ability.
vi. They have increased risk for blood clots.

4. **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 grow slowly than their colleague during growing time.
   ii. They are short stature (height) without puberty and menstrual cycle.
   iii. They have deformed (misshapen) ears, receding lower jaw.
   iv. They have webbed neck and broad chest.
   v. They have multiple tiny coloured spots on skin.
   vi. They have poorly developed ovaries and underdeveloped breasts.
   vii. They are sterile females
   viii. They possess many male characters like heavy neck muscles and narrow hips.

5. **Autism:** Autism (Autism spectrum disorder) is a neurological and developmental disorder. Person with autism may have problems with social interaction and communication skills. Although autism can be diagnosed at any age, it generally appears in the first two years of life.

   Symptoms:

   i. They fail to respond to their name or appear.
   ii. They resist cuddling and holding. They seem to prefer playing alone, retreating into their own world.
   iii. They have poor eye contact and lack facial expression.
   iv. They don't speak, delayed speech or loses ability to say word or sentences.
   v. They can't start a conversation.
   vi. They speak with an abnormal tone or rhythm and may use a singsong voice or robot like speech.
   vii. They repeat words or phrases.
   viii. They don't express emotions or feelings and unaware of others' feelings.
   ix. They can't recognize nonverbal signs.
   x. They may harm themselves, such as biting or head-banging.
   xi. They are unusually sensitive to light or sound. They may be indifferent to pain or temperature.
   xii. They have specific food preferences, such as eating only few foods or refusing foods with a certain texture.

6. **Albinism:** It is a genetic disability marked by few or none of the melanin pigment in the skin, hair and/or eyes. 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. They may develop freckles, moles with or without colour and sunburn when exposed to sun.
iii. It causes poor vision, but they show normal life.
iv. They are extremely sensitive to bright light (photophobia).
v. They have uncommon head position or head posture, such as tilting the head to reduce eye movements and see better.
vi. Eyes can't look at the same direction at the same time.

7. **Alzheimer's disease:** It is the most common cause of dementia. It is the biological process that begins with the appearance of a buildup of proteins in the form of amyloid planques and neurofibrillary tangles in the brain. This causes brain cells to die over time and the brain to shrink.
   Early symptom of this disease is forgetting recent events or conversations.
   Symptoms:
   i. Memory loss is the major symptom of this disease.
   ii. They show poor judgement or find it harder to make decisions.
   iii. They have problems with speech or language, disturb sleep, and change in mood.
   iv. They may need help for eating, washing, getting dresses, using the toilet, etc.
   v. They show difficulty in eating and swallowing.
   vi. They can't changing position or moving without assistance.
   vii. They may weight loss, unintentional passing of urine or stool, gradual loss of speech.

8. **Haemophilia:** It is a rare genetic disorder in which the blood doesn't clot because of lacking blood clotting proteins. The medication is regular replacement of the specific clotting factor that is reduced.
   Symptoms:
   i. Excessive or heavy bleeding from cuts or injuries.
   ii. Many large or deep bruises (discolouration).
   iii. It may pain, swelling or tightness in joints, blood in urine or stool.
   iv. Unusual bleeding after vaccination, nose bleed without any cause.
   v. Painful, prolonged headache, repeated vomiting, sleepiness or tiredness.