4

HEREDITY AND EVOLUTION

INTRODUCTION

The branch of biology that studies the process of heredity and variations is called genetics. Heredity is the ability of a living organism to pass on information about itself to its offspring concerning its physical and chemical makeup. When an organism reproduces asexually, a one-parent organism produces genetically identical offspring. The offspring are identical to the parent because all the genes from that parent end up in the offspring. When an organism reproduces sexually, genes from both parents help determine the characteristics of the offspring. In both of these types of reproduction, the offspring tend to resemble the parent or parents because of this transfer of genes.

However, we all know that there are subtle, recognizable differences between parents offspring. Impressed upon each individual is a unique genetic signature. These differences, are called variations.

Some variation is evident to all of us. Groups of people from different parts of the world are often quite different in appearance. Within any of these groups, individuals of one family often differ greatly from those of another. Even your brothers and sisters do not resemble you exactly, unless you have an identical twin. Nor are we human beings unique in this respect. Great differences in appearance often exist within other species. For example, there is a bewildering array of varieties and breeds of dogs, of every size and form imaginable, yet all are still dogs, able to interbreed and produce puppies.

Variation is not surprising in itself. Differences in diet during development can have marked effects on adult appearance, as can variation in the environments that different individuals experience. Many arctic mammals, for example, develop white fur when they are exposed to the cold of winter and dark fur during the warm summer months. The remarkable property of some of the pattern of variation that we can observe a property that has always fascinated and puzzled us is that some of the differences between individuals are inherited, passed down from parent to offspring.

GENE

Genes are working subunits of DNA. DNA is a vast chemical information database that carries the complete set of instructions for making all the proteins a cell will ever need. Each gene contains a particular set of instructions, usually coding for a particular protein.

DNA exists as two long, paired strands spiraled into the famous double helix. Each strand is made up of millions of chemical building blocks called bases. While there are only four different chemical bases in DNA (adenine, thymine, cytosine, and guanine), the order in which the bases occur determines the information available, much as specific letters of the alphabet combine to form words and sentences.

DNA resides in the core, or nucleus, of each of the body's trillions of cells. Every human cell (with the exception of mature red blood cells, which have no nucleus) contains the same DNA. Each cell has 46 molecules of double-stranded DNA. Each molecule is made up of 50 to 250 million bases housed in a chromosome.

The DNA in each chromosome constitutes many genes (as well as vast stretches of noncoding DNA, the function of which is unknown). A gene is any given segment along the DNA that encodes instructions that allow a cell to produce a specific product typically, a protein such as an enzyme- that initiates one specific action. There are between 50,000 and 100,000 genes, and every gene is made up of thousands, even hundreds of thousands, of chemical bases.

Human cells contain two sets of chromosomes, one set inherited from the mother and one from the father. (Mature sperm and egg cells carry a single set of chromosomes.) Each set has 23 single chromosomes - 22 autosomes and an X or Y sex chromosome. (Females inherit an X from each parent, while males get an X from the mother and a Y from the father.)

CHROMOSOMES

- Rod like bodies found in every cell (except red blood cells) in the body.
- Contain the genes or genetic code that determine various traits (the physical characteristics of an animal).
- There are many genes on each chromosome.

Base pair : Two bases which form a "rung of the DNA ladder." A DNA nucleotide is made of a molecule of sugar, a molecule of phosphoric acid, and a molecule called a base. The bases are the "letters" that spell out the genetic code. In DNA, the code letters are A, T, G and C, which stand for the chemicals adenine, thymine, guanine, and cytosine, respectively. In base pairing, adenine always pairs with thymine, and guanine always pairs with cytosine.

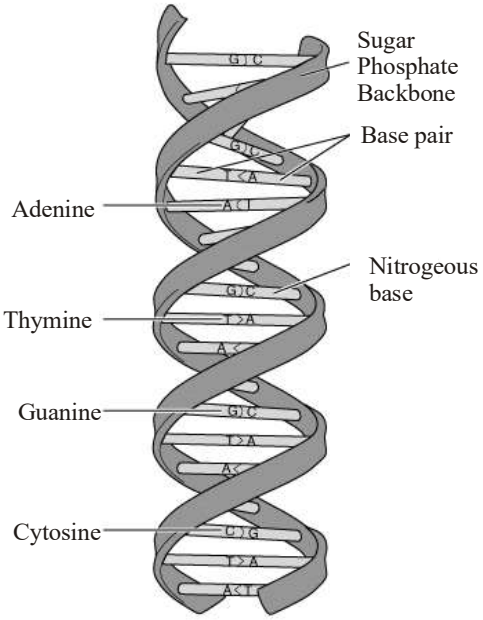


Figure : DNA structure

Genotype and phenotype: The particular set of genes that an individual acquires from its parents is known as its genotype. This remains constant throughout life and is generally unchanged by environmental events. However, the environment does affect how every organism develops and functions. For example, a person with tall parents likely has a genotype that encodes genes necessary for becoming tall as well; however, if that person lacks proper nutrition while growing or suffers from hormonal defects, he or she may not reach his/her full potential height. The form taken by some traits (such as tallness) in specific individuals is called a phenotype. Phenotype is determined in part by the genotype of the individual and in part by the environment to which they are exposed. Thus, the observable properties of an organism are produced by the combination of the genotype and its interaction with the environment. This means that a single genotype can produce different phenotypes, depending on the environment in which the organism develops.

For instance, identical twins have the same genotype, but could develop different phenotypes for certain traits if they were separated at birth and grew up in very different environments. Their adult heights might differ depending on their nutrition while growing up and other environmental factors. Phenotypic traits are not necessarily genetic. In simple words Exact DNA sequence of an individual is its genotype. The collection of all observable and measurable traits of that individual is phenotype.

On the other hand, the same phenotype can be produced by different genotypes, depending on the environment. For example, certain people all living in the same region with a prevalence of a specific disease may all become resistant even though they have different genotypes. The way their bodies acquire resistance may differ slightly due to genetic differences, but the overall observable trait of resistance is similar.

All humans have very similar sets of genes that specify in a general sense how we develop and function. However, there can be many different forms of a given gene; for example, the genes for eye colour are slightly different between people with brown eyes and those with blue eyes. (Note: eye colour is actually determined by a combination of several genes, but for the purpose of this example we will assume it is controlled by a single

gene.). These slightly different forms of the same gene are called alleles. Every person has two alleles for any given gene, one inherited from their mother and another from their father. In some cases, the alleles are identical, but they can also be slightly different.

Homozygous : Possessing two identical forms of a particular gene, one inherited from each parent.

Dominant: A gene that almost always results in a specific physical characteristic, for example, a disease, even though the patient's genome possesses only one copy. With a dominant gene, the chance of passing on the gene (and therefore the disease) to children is 50-50 in each pregnancy

Recessive : A genetic disorder that appears only in patients who have received two copies of a mutant gene, one from each parent.

First and second filial generation : The generation produced by the hybridisation between parents is called first generation. It is abbreviated as F_1. Further the offsprings produced by the cross of two hybrid plants or F_1 plants is called second filial generation. They are abbreviated as F_2.

Monohybrid cross : A monohybrid cross involves a single characteristic that is controlled by one gene with two or more alleles. The most commonly used example for monohybrid crosses is Mendel's experiments on pea plants (Pisum sativum) involving tall and dwarf specimens.

The letter T represents the dominant allele (tall) and the letter t represents the recessive allele (dwarf). A homozygous dominant specimen would have a genotype of TT, whereas a homozygous recessive plant would be tt. Being homozygous, the gametes of each parent would be the same (T or t, depending on the specimen) and therefore the cross would be written as follows:

	t	**t**
T	Tt	Tt
T	Tt	Tt

The resulting offspring are all heterozygous in genotype (Tt) but, since the dominant allele expresses the tall phenotype, all appear identical to the homozygous dominant parent (TT).

If these heterozygous offspring were interbred, the result would be somewhat different:

	T	**t**
T	TT	Tt
t	Tt	tt

The resulting offspring include two heterozygous specimens and two homozygous (dominant and recessive), although the phenotype ratio is 3:1 in terms of tall:dwarf.

In this way, it is simple to calculate that TT X Tt would result in two dominant homozygous (TT) and two heterozygous (Tt), and that Tt X tt would result in two recessive homozygous (tt) and two heterozygous (Tt).

DIHYBRID CROSS

Dihybrid cross is a cross between individuals that involves two pairs of contrasting traits. Predicting the results of a dihybrid cross is more complicated than predicting the results of a monohybrid cross. All possible combinations of the four alleles from each parent must be considered.

A dihybrid cross can be treated as two separate monohybrid crosses.

- In monohybrid crosses, to know if a dominant trait is homozygous (RR) or heterozygous (Rr) it is necessary to carry out a test cross
- This is done with a homozygous recessive (rr) individual
- The same is true for a dihybrid cross where the test cross is made with an individual which is homozygous recessive for both characters (rryy)

Simple dihybrid cross :

	AB	Ab	aB	ab
AB	AABB	AABb	AaBB	AaBb
Ab	AABb	AAbb	AaBb	Aabb
aB	AaBB	AaBb	aaBB	aaBb
ab	AaBb	Aabb	aaBb	aabb

Basic terms :

Term	Definition
Allele	An alternate form of a gene
Dihybrid	An individual who is heterozygous for two particular genes
Dominant	An allele that masks the expression of another allele
F_1	The first filial generation; offspring
F_2	The second filial generation; offspring of offspring
Genotype	The allele combination in an individual
Heterozygous	Possessing different alleles of a gene
Homozygous	Possessing identical alleles of a gene
Independent assortment	Mendel's second law; a gene on one chromosome does not influence the inheritance of a gene on a different (nonhomologous) chromosome because meiosis packages chromosomes randomly into gametes
Monohybrid	An individual heterozygous for a particular gene
Mutant	A phenotype or allele resulting from a change (mutation) in a gene
Mutation	A change in a gene
P_1	The parental generation
Phenotype	The observable expression of an allele combination
Recessive	An allele whose expression is masked by another allele
Segregation	Mendel's first law; alleles of a gene separate into equal numbers of gametes
Sex-linked	A gene located on the X chromosome or a trait that results from the activity of a gene on the X chromosome
Wild type	The most common phenotype or allele for a gene in a population

INHERENT AND NON-INHERENT CHARACTERS

The general meaning of trait is character. Several traits are found in an animal or a plant. All these characters constitute the identity of the organism. We often listen peculiar statements for different individuals like 'that dwarf, curly haired and blue eyed gentleman' or 'that tall, bald headed and lame man'. The characters described in the above statements are termed traits in genetics. The traits found in an organism can be divided in two types. First, those characters or traits which are received by an individual from the parents and which are present since his birth. Such traits are called inherent traits which inherit in next generation. In above examples the characters like dwarfness or tallness and blue eyes etc. are inherent characters. Second, those characters which are acquired by an individual in his own life time by way of environmental or health reasons. Such characters are not inherited in the future generations.. These are called non-inherent traits. The development of lameness in a man is an example of such a character. Similarly muscularity of a wrestler is also a trait of non-inherent category. Several diseases in man are examples of inherent character like sickle cell anemia, hemophilia, mangolism etc.

Organisms differ in many traits, or characters. A character is a feature of an organism such as its height, flower color, or the chemical structure of a protein. Many characters are determined at least in part by genes, which are individual units of genetic information for specific characters. The gene is the basic unit of inheritance.

The body cells of most plants and animals are diploid-that is, they contain two copies of each chromosome type. Therefore, they contain two copies of each gene, one inherited from each parent. These two copies are not necessarily identical, however. Such alternative versions of a gene are called alleles. The different alleles of a gene are often denoted by uppercase and lowercase letters-for example, A and a. No individual has more than two alleles for a particular gene, but among a large number of individuals of the same species a single gene can be represented by many more than two alleles. An individual that carries two copies of the same allele, such as an AA or an aa individual, is called a homozygote. An individual that carries one copy of each of two different alleles, such as an Aa individual, is called a heterozygote.

The genetic makeup of an organism is called its genotype; for example, a heterozygote has genotype Aa. The phenotype of an organism is its observable physical characteristics. Aspects of an organism's phenotype include its appearance (for example, flower color), behavior (for example, the courtship display of a bird), and biochemistry (for example, the amount of a gene's protein product in the body). Two individuals with the same phenotype can have different genotypes.

An allele that determines the phenotype of an organism even when it is paired with a different allele is referred to as a dominant allele. Dominant alleles are often denoted by uppercase letters, such as A. An allele that does not express its phenotypic effect when paired with a dominant allele is said to be recessive. Recessive alleles are often denoted by lowercase letters, such as a.

A genetic cross, or cross for short, is a controlled mating experiment performed to examine the inheritance of a particular character. "Cross" can also be used as a verb, as in "individuals of genotype AA were crossed with individuals of genotype aa." The parent generation of a genetic cross is called the P generation. The first generation of offspring in a genetic cross is called the F, generation ("F" is for "filial," a word that refers to a son or daughter). The second generation of a cross is called the F_2 generation.

Two aspects to heredity :

Mendelian Genetics : Quantitative analysis of inheritance of morphological characters in organisms experiments which established the basic principles & laws that predict the pattern of inheritance from generation to generation

Molecular Genetics : Physical properties of molecules (DNA & RNA) as they effect patterns of inheritance.
Central Dogma of Molecular Biology : DNA $\rightarrow$ transcription $\rightarrow$ RNA $\rightarrow$ translation $\rightarrow$ Protein
You will learn about molecular genetics in higher classes.

Mendel Experiment :

Mendel selected seven pairs of characters which could easily be seen in the garden pea. They were :
(i) Length of stem – Long and short
(ii) Position of bud – Terminal and oxiliary
(iii) Texture of seed coat – Smooth and wrinkled
(iv) Colour of seed – Yellow and green
(v) Colour of flower – Red and white
(vi) Shape of pod – Inflated or deflated

Why he selected Garden pea for his experiment :

(i) It is small plant. Large number of plants could easily be grown in his small garden.
(ii) It is an annul and flower within 60-90 days. Two crops in a year ihence mere generations could in studied in a short period.
(iii) Possess sharply distinguished characters. Which could easily be observed without any optical aid ?
(iv) It is self pollinated plant and male and female reproductive organs are protected in boat shaped structure – the Keel. Hence it is easy to carry out artificial pollination.

Experiment : Mendel selected two strains of garden peas – the long plant and dwarf plant. He subjected each of these for self pollination and procured seeds. On sowing seeds, he found out that tall ones gave tall progeny and dwarf ones gave dwarf progeny. He repeated his process for a number of generation to ensure that the two strain should be pure for characters – Tallness and dwarfness. He called these two as plants true for characters –tallness and dwarfness. He treated each one as parents in his experiments of breeding of peas. He called this generation as parent generation(P). He obtained seeds by artificial pollination and by sowing these seeds he obtained first offspring generation to which he named.

First Filial generation (F_1) since F_1 was obtained by crossing parents with two different traits–tall and dwarf, he called F_1 generation as hybrid generation. In F_1 generation all plants were tall.

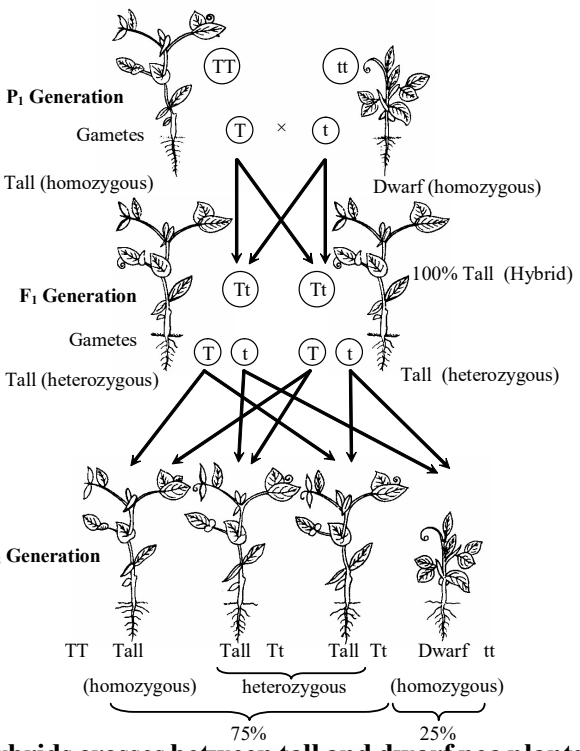


Figure : Mendel's monohybrids crosses between tall and dwarf pea plants

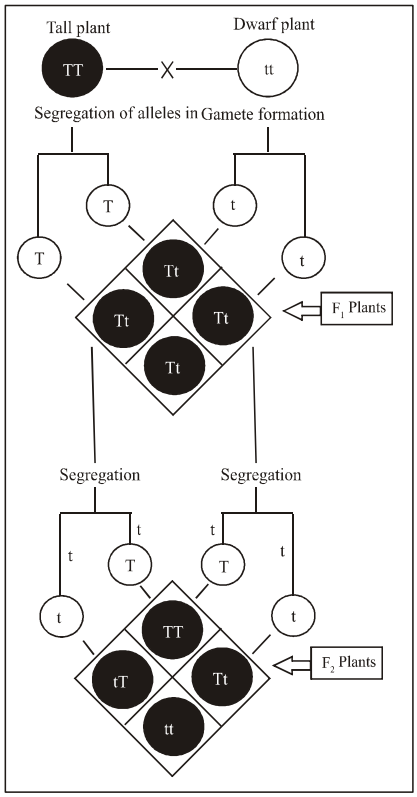
Obviously dwarfness did not express itself in F_1. The trait that appear in F_1 (tallness in this example) is termed as dominant. The trait which is not expressed in F_1 is called recessive (dwarfness in this experiment)

On the basis of such experiments Mendel formulated the first law of Dominance and Recessiveness.

LAW OF DOMINANCE AND RECESSIVENESS

When parents true breeding for contrasting traits are crossed only the dominant trait appear in first generation or hybrid generation. What happened to the recessive trait (dwarfness in the example) ? In order to find out the solution of this problem Mendel did further experiment. He crossed the plants of F_1 generation among themselves (inbreeding) and also subjected them to self pollination and obtained seeds. The plants raised by these seeds – represented (F_2) second filial generation. In this generation he got dwarf plants as well as tall plants but in fixed proportion of 3 tall : 1 Dwarf.

On the basis of such experiments he formulated the second law of seggregation. When hybrids are selfed or inbred the dominant and recessive traits seggregate and recessive character appears in F_2 of the total offspring.



In this experiment the ratio 3 : 1 is based on traits that appeared or physically expressed. The ratio of physically expressed or observed trait is called phenotypic ratio.

Mendel regarded that certain particles which he called as 'factors' control the characters. These factors are passed on from parents to offspring by eggs and sperm, through sexual reproduction. The discovery of meiosis and resembles of chromosome behaviour during meiosis lead to the concept that Mendelian factors were infact 'genes' that are located on chromosomes in a linear fashion. In diploid condition we know that chromosome are found in pairs. Mendel's factor also occur in pair. By meiosis only one member of pair of chromosome goes to gamete, so is the 'factor'.

How the gene or factor behaves during inheritance can be now well understood. The allele for dominant trait tallness shown as capital 'T' and recessive trait dwarfness as small 't'. If the checker square the gene situation given in circle. The phenotypic ratio as explained above is 3 tall : 1 dwarf. If you look at the exact situation of 'genes' you will find 1 pure tall : 2 hybrid tall : 1 dwarf. That is ratio with respect to gene situation is 1 : 2 : 1. Hence this ratio is called genotypic ratio.

In an individual when two alleles for trait are of the same type it is called homozygous condition (for example TT or tt) when in an individual two alleles are contrasting type, it is called homozygous condition (for example Tt). Hybrid plants are always heterozygous for a character while true breeding plants for a character as always homozygous for that character.

Mendel did experiments on inheritance of two character at a time. In one such experiment he studied the inheritance of size of the plant and colour of flower. Tallness dominant over dwarfness and red colour of flower dominant over white colour. He selected two true breeding parents Tall with red flower and dwarf with white flowers. By artificial crossing he obtained seed. By raising these seeds he obtained F_1 or hybrid generation. All plants showed dominants, they were tall with red flowers. By self pollination, the seeds obtained from F_1 plants were sown. The F_2 generation has two new types of combination in addition to two parental types. Thus in F_2 parental types were tall and red flower, Dwarf and white flowered.

The new types were Tall and white, dwarf and red flowered. The ratio of the 4 types was 9 tall, red : 3 tall white : 3 Dwarf red, 1 dwarf white. This indicates that non-contrasting characters such as size and colour are inherited independent of one another. On this basis Mendel formulated the 3rd law known as Law of independent assortment. According to this law, non-contrasting characters are inherited independent of one another.

In the above example the inheritance two non-contrasting characters (size and colour) were studied hence it is dihybrid and ratio 9 : 3 : 3 : 1 is dihybrid phenotypic ratio. In previous example of inheritance of size only one character was that is size was considered and ratio 3 : 1 and 1 : 2 : 1 represents monohybrid ratios. The results of Mendel's F_1 crosses for seven characters in pea plants.

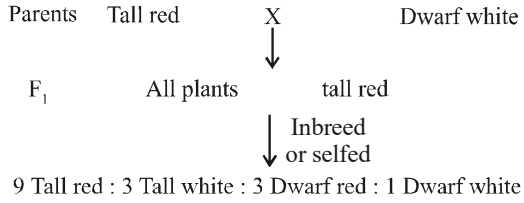


Figure : Schematic diagram of a dihybrid cross.

SEX DETERMINATION

In lower invertebrates such as amoeba, earthworm the male and female cannot be distinguished on the basis of external characters. In higher animals such as cattles, apes and man the two can easily be distinguished. It was found out that characters maleness and femaleness are related to chromosomes This fact was first discovered by T.H. Morgan in fruit fly (Drosophila melanofaster). In this fly 2n-8, that is there are 4 pairs of homologous chromosomes. All the four pairs of fruit fly chromosomes have been identified and numbered. The II, III and IV pairs of chromosomes are identical in the cells of both male and female fly.

They are called autosomes (non-sex chromosomes). First pair is the sex-chromosomes. In female both partners of pair are rod shaped and called X-chromosome. In male fly there is one rod shaped X-chromosome and the other is J shaped or hook shape called Y-chromosome.

In fruit fly it was found that sperm determines the sex of the offspring. All eggs are alike carrying one X-chromosome but sperm are of two types. One type having X-chromosome and other carrying Y-chromosomes. When an X carrying sperm fertilizes an egg the zygote is XX, develop into a female. XX carrying sperm produces XY zygote develop into a male.

Normally the fruit by possess red eyes. In one of the morgan's culture their appeared flies having white-eyes. When a white eyed male crossed with red eyed female, all the offsprings were red eyed. He crossed this offsprings with one another again (in breeding) he got white eyed flies and red eyed flies. He observed that all white eyed flies were males. Thus he could establish that the gene for eye colour is located an X-chromosomes. So it is a trait linked with sex chromosomes.

Such a mechanism of sex determination have also been discovered in other animals like birds. In bird males have identical sex chromosomes (ZZ) while the females have unlike partners in sex homologue (ZW).

In man male and female are easily distinguished. Here also maleness and femaleness depends on difference in chromosomes. Like fruit fly the human female basically have XX chromosomes and human male have XY. The 2-n number in man is 46 that is 23 homologues pairs. 22 pairs are identical in man and woman while one pais is XX females and XY in males. When haploid gametes (sperms) are formed after meiosis, they are of two types, 50% will be (22+X) and rest 50% will be 22+ Y. In female all egg will be of one type (22 + X). When an egg (22 + X) gets fertilized by a sperm of (22 + X) types. The Zygote (44 + XX) will develop into a female child. If egg (22 + X) gets fertilized by sperm (22+Y) type than Zygote (44 + XY) will develop into a male. Thus in man it is the sperms which determines the sex of child and there are 50 : 50 chances of having a son or daughter.

EVOLUTION

The word evolution means 'an unrolling'. When word evolution applied to living organisms, the intention is to convey the concept that plants and animals of today have 'unrolled' – that is developed from those of the past. The present day species are different from the ancestral types.

Modern concepts of evolution is based on the following –

1. Plants and animals have changed in the past, they are still changing and they will continue to change in future.
2. The changes that have occurred in past ages have gradually given rise to different groups of organisms, many of them are still living.
3. The evolution of new groups is gradual and may need millions of years. Therefore from an species, evolution of a new species cannot be observed in one's life time.
4. Present day species are more complex in structure and functions than the past. Thus simpler forms have given rise to more complex forms during evolution. Hence simpler forms are called primitive while evolved forms called advance or specialized. However in some cases the evolution is backward or retrogressive. That is simpler forms derived from complex forms as is evident in many parasitic species like liver fluke, ascaris etc. Where it is presumed that there ancestors were more complex.

These underlying concepts are based on evidences drawn from different fields like taxonomy, comparative anatomy, embryology, paleontology, comparative-physiology, cytogenetics, breeding and geographical distribution.

Evidences from Paleontology : Paleontology is the study of fossils. 'Fossil' literary means 'dug from earth'. In Biology fossil means 'any remains or traces of a once living organism'.

These remains may be a part or its impression or the organ or body surrounded and permeated by sand stone and turned into rock. Fossils are found accidently or they are systematically hunted and excavated by paleontologists. In laboratories, they are freed from clinging rocks, cleaned, sliced and microscopically examined. Jumbled parts of ancient living things are then arranged and the entire organism is reconstructed. Most existing fossils are still buried in the ancient sedimentary rocks. Only a small number have been found and studied so far.