

**S.Y.B.Sc. Life Sciences Semester IV (CBSGS)**

**Paper III**

**Dated: 2<sup>nd</sup> May 2019**

**Q.P.Code: 34687**

**Answer Key**

**Q.1. Do As Directed:**

**A.Define :** **(07)**

**1.Kin Selection:** Natural selection in which an apparently disadvantageous characteristic (especially altruistic behaviour) increases in the population due to increased survival of individuals genetically related to those possessing the characteristic.

**2.Communication:** Communication is the act of conveying meanings from one entity or group to another through the use of mutually understood signs, symbols, and semiotic rules.

**3.Sympatric Speciation:** Sympatric speciation is the evolution of a new species from a surviving ancestral species while both continue to inhabit the same geographic region.

**4.Cultural Evolution:** Cultural evolution is an evolutionary theory of social change. It follows from the definition of culture as "information capable of affecting individuals' behavior that they acquire from other members of their species through teaching, imitation and other forms of social transmission".

**5.Reciprocal Altruism:** **Reciprocal altruism** is a behaviour whereby an organism acts in a manner that temporarily reduces its fitness while increasing another organism's fitness, with the expectation that the other organism will act in a similar manner at a later time.

**6.Evolution:** Evolution is the process of heritable change in populations of organisms over multiple generations.

**7.Analogy:** A comparison between one thing and another, typically for the purpose of explanation or clarification.

**Q.1 B. Match the columns:** **(07)**

**a- v, b-vi, c-i, d-iv, e-iii, f-vii, g-ii**

**Q. 1 C. State whether True or False:** **(06)**

**1. True 2.True 3. False 4. False 5. False 6. False**

**Q.2.A. Answer any one of the following:** **(10)**

**1.What are the selective social and behavioral characteristics of hunting societies?**

A hunter-gatherer is a human living in a society in which most or all food is obtained by foraging (collecting wild plants and pursuing wild animals). Hunter-gatherer societies stand in contrast to agricultural societies, which rely mainly on domesticated species.

Hunting and gathering was humanity's first and most successful adaptation, occupying at least 90 percent of human history. Following the invention of agriculture, hunter-gatherers

who did not change have been displaced or conquered by farming or pastoralist groups in most parts of the world. **Hunting and gathering societies** survive by hunting game and gathering edible plants. Until about 12,000 years ago, all societies were hunting and gathering societies.

There are five basic characteristics of hunting and gathering societies:

1. The primary institution is the family, which decides how food is to be shared and how children are to be socialized, and which provides for the protection of its members.
2. They tend to be small, with fewer than fifty members.
3. They tend to be nomadic, moving to new areas when the current food supply in a given area has been exhausted.
4. Members display a high level of interdependence.
5. Labor division is based on sex: men hunt, and women gather.

The **first social revolution**—the domestication of plants and animals—led to the birth of the horticultural and pastoral societies.

Hunting and gathering societies are slowly disappearing, as the encroachment of civilization destroys the land they depend on. The Pygmies in Africa are one of the few remaining such societies.

Hunter-gatherers tend to have an egalitarian social ethos, although settled hunter-gatherers (for example, those inhabiting the Northwest Coast of North America) are an exception to this rule. Nearly all African hunter-gatherers are egalitarian, with women roughly as influential and powerful as men. Karl Marx defined this socio-economic system as primitive communism.

The egalitarianism typical of human hunters and gatherers is never total, but is striking when viewed in an evolutionary context. One of humanity's two closest primate relatives, chimpanzees, are anything but egalitarian, forming themselves into hierarchies that are often dominated by an alpha male. So great is the contrast with human hunter-gatherers that it is widely argued by palaeoanthropologists that resistance to being dominated was a key factor driving the evolutionary emergence of human consciousness, language, kinship and social organization.

Anthropologists maintain that hunter/gatherers don't have permanent leaders; instead, the person taking the initiative at any one time depends on the task being performed. In addition to social and economic equality in hunter-gatherer societies, there is often, though not always, sexual parity as well. Hunter-gatherers are often grouped together based on kinship and band (or tribe) membership. Postmarital residence among hunter-gatherers tends to be matrilineal, at least initially. Young mothers can enjoy childcare support from their own mothers, who continue living nearby in the same camp. The systems of kinship and descent among human hunter-gatherers were relatively flexible, although there is evidence that early human kinship in general tended to be matrilineal. One common arrangement is the sexual division of labour, with women doing most of the gathering, while men concentrate on big game hunting. In all hunter-gatherer societies, women appreciate the meat brought back to camp by men.

## **2. Discuss the concept of isolation mechanism.**

The mechanisms of reproductive isolation are a collection of evolutionary mechanisms, behaviors and physiological processes critical for speciation. They prevent members of

different species from producing offspring, or ensure that any offspring are sterile. These barriers maintain the integrity of a species by reducing gene flow between related species.

The mechanisms of reproductive isolation have been classified in a number of ways. Zoologist Ernst Mayr classified the mechanisms of reproductive isolation in two broad categories: pre-zygotic for those that act before fertilization (or before mating in the case of animals) and post-zygotic for those that act after it. The mechanisms are genetically controlled and can appear in species whose geographic distributions overlap (sympatric speciation) or are separate (allopatric speciation).

Pre-zygotic isolation mechanisms are the most economic in terms of the natural selection of a population, as resources are not wasted on the production of a descendant that is weak, non-viable or sterile. These mechanisms include physiological or systemic barriers to fertilization.

#### Temporal or habitat isolation

The Central Valley in California prevents two the salamander population from interacting with each other which is a habitat isolation. After many generations the two salamander gene pools will become mutated caused by natural selection. The mutation will change the DNA sequence of the two populations enough that the salamander populations can no longer successfully breed between each other making the populations of salamander become classified as different species.

Any of the factors that prevent potentially fertile individuals from meeting will reproductively isolate the members of distinct species. The types of barriers that can cause this isolation include: different habitats, physical barriers, and a difference in the time of sexual maturity or flowering. An example of the ecological or habitat differences that impede the meeting of potential pairs occurs in two fish species of the family Gasterosteidae (sticklebacks). One species lives all year round in fresh water, mainly in small streams. The other species lives in the sea during winter, but in spring and summer individuals migrate to river estuaries to reproduce. The members of the two populations are reproductively isolated due to their adaptations to distinct salt concentrations. An example of reproductive isolation due to differences in the mating season are found in the toad species *Bufo americanus* and *Bufo fowleri*. The members of these species can be successfully crossed in the laboratory producing healthy, fertile hybrids. However, mating does not occur in the wild even though the geographical distribution of the two species overlaps. The reason for the absence of inter-species mating is that *B. americanus* mates in early summer and *B. fowleri* in late summer. Certain plant species, such as *Tradescantia canaliculata* and *T. subaspera*, are sympatric throughout their geographic distribution, yet they are reproductively isolated as they flower at different times of the year. In addition, one species grows in sunny areas and the other in deeply shaded areas.

#### Behavioral isolation

The different mating rituals of animal species creates extremely powerful reproductive barriers, termed sexual or behavior isolation, that isolate apparently similar species in the majority of the groups of the animal kingdom. In dioecious species, males and females have to search for a partner, be in proximity to each other, carry out the complex mating rituals and finally copulate or release their gametes into the environment in order to breed.

The songs of birds, insects and many other animals are part of a ritual to attract potential partners of their own species. The song presents specific patterns recognizable only by members of the same species, and therefore represents a mechanism of reproductive isolation. This recording is the song of a species of cicada, recorded in New Zealand.

Mating dances, the songs of males to attract females or the mutual grooming of pairs, are all examples of typical courtship behavior that allows both recognition and reproductive isolation. This is because each of the stages of courtship depend on the behavior of the partner. The male will only move onto the second stage of the exhibition if the female shows certain responses in her behavior. He will only pass onto the third stage when she displays a second key behavior. The behaviors of both interlink, are synchronized in time and lead finally to copulation or the liberation of gametes into the environment. No animal that is not physiologically suitable for fertilization can complete this demanding chain of behavior. In fact, the smallest difference in the courting patterns of two species is enough to prevent mating (for example, a specific song pattern acts as an isolation mechanism in distinct species of grasshopper of the genus *Chorthippus*). Even where there are minimal morphological differences between species, differences in behavior can be enough to prevent mating. For example, *Drosophila melanogaster* and *D. simulans* which are considered twin species due to their morphological similarity, do not mate even if they are kept together in a laboratory. *Drosophila ananassae* and *D. pallidosa* are twin species from Melanesia. In the wild they rarely produce hybrids, although in the laboratory it is possible to produce fertile offspring. Studies of their sexual behavior show that the males court the females of both species but the females show a marked preference for mating with males of their own species. A different regulator region has been found on Chromosome II of both species that affects the selection behavior of the females.

Pheromones play an important role in the sexual isolation of insect species. These compounds serve to identify individuals of the same species and of the same or different sex. Evaporated molecules of volatile pheromones can serve as a wide-reaching chemical signal. In other cases, pheromones may be detected only at a short distance or by contact.

In species of the *melanogaster* group of *Drosophila*, the pheromones of the females are mixtures of different compounds, there is a clear dimorphism in the type and/or quantity of compounds present for each sex. In addition, there are differences in the quantity and quality of constituent compounds between related species, it is assumed that the pheromones serve to distinguish between individuals of each species. An example of the role of pheromones in sexual isolation is found in 'corn borers' in the genus *Ostrinia*. There are two twin species in Europe that occasionally cross. The females of both species produce pheromones that contain a volatile compound which has two isomers, E and Z; 99% of the compound produced by the females of one species is in the E isomer form, while the females of the other produce 99% isomer Z. The production of the compound is controlled by just one locus and the interspecific hybrid produces an equal mix of the two isomers. The males, for their part, almost exclusively detect the isomer emitted by the females of their species, such that the hybridization although possible is scarce. The perception of the males is controlled by one gene, distinct from the one for the production of isomers, the heterozygous males show a moderate response to the odour of either type. In this case, just 2 'loci' produce the effect of ethological isolation between species that are genetically very similar.

Sexual isolation between two species can be asymmetrical. This can happen when the mating that produces descendants only allows one of the two species to function as the female progenitor and the other as the male, while the reciprocal cross does not occur. For instance, half of the wolves tested in the Great Lakes area of America show mitochondrial DNA sequences of coyotes, while mitochondrial DNA from wolves is never found in coyote populations. This probably reflects an asymmetry in inter-species mating due to the difference in size of the two species as male wolves take advantage of their greater size in order to mate with female coyotes, while female wolves and male coyotes do not mate.

Mechanical isolation

The flowers of many species of Angiosperm have evolved to attract and reward a single or a few pollinator species (insects, birds, mammals). Their wide diversity of form, colour, fragrance and presence of nectar is, in many cases, the result of coevolution with the pollinator species. This dependency on its pollinator species also acts as a reproductive isolation barrier.

Mating pairs may not be able to couple successfully if their genitals are not compatible. The relationship between the reproductive isolation of species and the form of their genital organs was signaled for the first time in 1844 by the French entomologist Léon Dufour. Insects' rigid carapaces act in a manner analogous to a lock and key, as they will only allow mating between individuals with complementary structures, that is, males and females of the same species (termed co-specifics).

Evolution has led to the development of genital organs with increasingly complex and divergent characteristics, which will cause mechanical isolation between species. Certain characteristics of the genital organs will often have converted them into mechanisms of isolation. However, numerous studies show that organs that are anatomically very different can be functionally compatible, indicating that other factors also determine the form of these complicated structures.

Mechanical isolation also occurs in plants and this is related to the adaptation and coevolution of each species in the attraction of a certain type of pollinator (where pollination is zoophilic) through a collection of morphophysiological characteristics of the flowers (called floral syndromes), in such a way that the transport of pollen to other species does not occur.

#### Gametic isolation

The synchronous spawning of many species of coral in marine reefs means that inter-species hybridization can take place as the gametes of hundreds of individuals of tens of species are liberated into the same water at the same time. Approximately a third of all the possible crosses between species are compatible, in the sense that the gametes will fuse and lead to individual hybrids. This hybridization apparently plays a fundamental role in the evolution of coral species. However, the other two-thirds of possible crosses are incompatible. It has been observed that in sea urchins of the genus *Strongylocentrotus* the concentration of spermatocytes that allow 100% fertilization of the ovules of the same species is only able to fertilize 1.5% of the ovules of other species. This inability to produce hybrid offspring, despite the fact that the gametes are found at the same time and in the same place, is due to a phenomenon known as gamete incompatibility, which is often found between marine invertebrates, and whose physiological causes are not fully understood.

In plants the pollen grains of a species can germinate in the stigma and grow in the style of other species. However, the growth of the pollen tubes may be detained at some point between the stigma and the ovules, in such a way that fertilization does not take place. This mechanism of reproductive isolation is common in the angiosperms and is called cross-incompatibility or incongruence. A relationship exists between self-incompatibility and the phenomenon of cross-incompatibility. In general crosses between individuals of a self-compatible species (SC) with individuals of a self-incompatible (SI) species give hybrid offspring. On the other hand, a reciprocal cross (SI x SC) will not produce offspring, because the pollen tubes will not reach the ovules. This is known as unilateral incompatibility, which also occurs when two SC or two SI species are crossed.

In coral reefs, gamete incompatibility prevents the formation of numerous inter-species hybrids.

#### Post-zygotic isolation

A number of mechanisms which act after fertilization preventing successful inter-population crossing are discussed below.

### Zygote mortality and non-viability of hybrids

A type of incompatibility that is found as often in plants as in animals occurs when the egg or ovule is fertilized but the zygote does not develop, or it develops and the resulting individual has a reduced viability. This is the case for crosses between species of the frog genus, where widely differing results are observed depending of the species involved. In some crosses there is no segmentation of the zygote (or it may be that the hybrid is extremely non-viable and changes occur from the first mitosis). In others, normal segmentation occurs in the blastula but gastrulation fails. Finally, in other crosses, the initial stages are normal but errors occur in the final phases of embryo development. This indicates differentiation of the embryo development genes (or gene complexes) in these species and these differences determine the non-viability of the hybrids.

Similar results are observed in mosquitos of the genus *Culex*, but the differences are seen between reciprocal crosses, from which it is concluded that the same effect occurs in the interaction between the genes of the cell nucleus (inherited from both parents) as occurs in the genes of the cytoplasmic organelles which are inherited solely from the female progenitor through the cytoplasm of the ovule.

In Angiosperms, the successful development of the embryo depends on the normal functioning of its endosperm.

The failure of endosperm development and its subsequent abortion has been observed in many interploidal crosses (that is, those between populations with a particular degree of intra or interspecific ploidy), and in certain crosses in species with the same level of ploidy. The collapse of the endosperm, and the subsequent abortion of the hybrid embryo is one of the most common post-fertilization reproductive isolation mechanism found in angiosperms.

### Hybrid sterility

Mules are hybrids with interspecific sterility.

A hybrid has normal viability but is typically deficient in terms of reproduction or is sterile. This is demonstrated by the mule and in many other well known hybrids. In all of these cases sterility is due to the interaction between the genes of the two species involved; to chromosomal imbalances due to the different number of chromosomes in the parent species; or to nucleus-cytoplasmic interactions such as in the case of *Culex* described above.

Hinnies and mules are hybrids resulting from a cross between a horse and a donkey or between a mare and a donkey, respectively. These animals are nearly always sterile due to the difference in the number of chromosomes between the two parent species. Both horses and donkeys belong to the genus *Equus*, but *Equus caballus* has 64 chromosomes, while *Equus asinus* only has 62. A cross will produce offspring (mule or hinny) with 63 chromosomes, that will not form pairs, which means that they do not divide in a balanced manner during meiosis. In the wild, the horses and donkeys ignore each other and do not cross. In order to obtain mules or hinnies it is necessary to train the progenitors to accept copulation between the species or create them through artificial insemination.

The sterility of many interspecific hybrids in angiosperms has been widely recognised and studied. Interspecific sterility of hybrids in plants has multiple possible causes. These may be genetic, related to the genomes, or the interaction between nuclear and cytoplasmic factors, as will be discussed in the corresponding section. Nevertheless, it is important to note that in plants, hybridization is a stimulus for the creation of new species – the contrary to the situation in animals. Although the hybrid may be sterile, it can continue to multiply in the wild by asexual reproduction, whether vegetative propagation or apomixis or the production of seeds. Indeed, interspecific hybridization can be associated with polyploidy and, in this way, the origin of new species that are called allopolyploids. *Rosa canina*, for example, is the

result of multiple hybridizations or there is a type of wheat that is an allohexaploid that contains the genomes of three different species.

#### Multiple mechanisms

In general, the barriers that separate species do not consist of just one mechanism. The twin species of *Drosophila*, *D. pseudoobscura* and *D. persimilis*, are isolated from each other by habitat (*persimilis* generally lives in colder regions at higher altitudes), by the timing of the mating season (*persimilis* is generally more active in the morning and *pseudoobscura* at night) and by behavior during mating (the females of both species prefer the males of their respective species). In this way, although the distribution of these species overlaps in wide areas of the west of the United States of America, these isolation mechanisms are sufficient to keep the species separated. Such that, only a few fertile females have been found amongst the other species among the thousands that have been analyzed. However, when hybrids are produced between both species, the gene flow between the two will continue to be impeded as the hybrid males are sterile. Also, and in contrast with the great vigor shown by the sterile males, the descendants of the backcrosses of the hybrid females with the parent species are weak and notoriously non-viable. This last mechanism restricts even more the genetic interchange between the two species of fly in the wild.

#### Hybrid sex: Haldane's rule

Haldane's rule states that when one of the two sexes is absent in interspecific hybrids between two specific species, then the sex that is not produced, is rare or is sterile is the heterozygous (or heterogametic) sex. In mammals, at least, there is growing evidence to suggest that this is due to high rates of mutation of the genes determining masculinity in the Y chromosome.

It has been suggested that Haldane's rule simply reflects the fact that the male sex is more sensitive than the female when the sex-determining genes are included in a hybrid genome. But there are also organisms in which the heterozygous sex is the female: birds and butterflies and the law is followed in these organisms. Therefore, it is not a problem related to sexual development, nor with the sex chromosomes. Haldane proposed that the stability of hybrid individual development requires the full gene complement of each parent species, so that the hybrid of the heterozygous sex is unbalanced (i.e. missing at least one chromosome from each of the parental species). For example, the hybrid male obtained by crossing *D. melanogaster* females with *D. simulans* males, which is non-viable, lacks the X chromosome of *D. simulans*.

#### Genetics

##### Pre-copulatory mechanisms in animals

The genetics of ethological isolation barriers will be discussed first. Pre-copulatory isolation occurs when the genes necessary for the sexual reproduction of one species differ from the equivalent genes of another species, such that if a male of species A and a female of species B are placed together they are unable to copulate. Study of the genetics involved in this reproductive barrier tries to identify the genes that govern distinct sexual behaviors in the two species. The males of *Drosophila melanogaster* and those of *D. simulans* conduct an elaborate courtship with their respective females, which are different for each species, but the differences between the species are more quantitative than qualitative. In fact the *simulans* males are able to hybridize with the *melanogaster* females. Although there are lines of the latter species that can easily cross there are others that are hardly able to. Using this difference, it is possible to assess the minimum number of genes involved in pre-copulatory isolation between the *melanogaster* and *simulans* species and their chromosomal location.

In experiments, flies of the *D. melanogaster* line, which hybridizes readily with *simulans*, were crossed with another line that it does not hybridize with, or rarely. The females of the segregated populations obtained by this cross were placed next to *simulans* males and the percentage of hybridization was recorded, which is a measure of the degree of reproductive

isolation. It was concluded from this experiment that 3 of the 8 chromosomes of the haploid complement of *D. melanogaster* carry at least one gene that affects isolation, such that substituting one chromosome from a line of low isolation with another of high isolation reduces the hybridization frequency. In addition, interactions between chromosomes are detected so that certain combinations of the chromosomes have a multiplying effect. Cross incompatibility or incongruence in plants is also determined by major genes that are not associated at the self-incompatibility S locus.

Post-copulation or fertilization mechanisms in animals

Reproductive isolation between species appears, in certain cases, a long time after fertilization and the formation of the zygote, as happens – for example – in the twin species *Drosophila pavani* and *D. gaucha*. The hybrids between both species are not sterile, in the sense that they produce viable gametes, ovules and spermatozoa. However, they cannot produce offspring as the sperm of the hybrid male do not survive in the semen receptors of the females, be they hybrids or from the parent lines. In the same way, the sperm of the males of the two parent species do not survive in the reproductive tract of the hybrid female. This type of post-copulatory isolation appears as the most efficient system for maintaining reproductive isolation in many species.

The development of a zygote into an adult is a complex and delicate process of interactions between genes and the environment that must be carried out precisely, and if there is any alteration in the usual process, caused by the absence of a necessary gene or the presence of a different one, it can arrest the normal development causing the non-viability of the hybrid or its sterility. It should be borne in mind that half of the chromosomes and genes of a hybrid are from one species and the other half come from the other. If the two species are genetically different, there is little possibility that the genes from both will act harmoniously in the hybrid. From this perspective, only a few genes would be required in order to bring about post copulatory isolation, as opposed to the situation described previously for pre-copulatory isolation.

In many species where pre-copulatory reproductive isolation does not exist, hybrids are produced but they are of only one sex. This is the case for the hybridization between females of *Drosophila simulans* and *Drosophila melanogaster* males: the hybridized females die early in their development so that only males are seen among the offspring. However, populations of *D. simulans* have been recorded with genes that permit the development of adult hybrid females, that is, the viability of the females is "rescued". It is assumed that the normal activity of these speciation genes is to "inhibit" the expression of the genes that allow the growth of the hybrid. There will also be regulator genes.

A number of these genes have been found in the *melanogaster* species group. The first to be discovered was "Lhr" (Lethal hybrid rescue) located in Chromosome II of *D. simulans*. This dominant allele allows the development of hybrid females from the cross between *simulans* females and *melanogaster* males. A different gene, also located on Chromosome II of *D. simulans* is "Shfr" that also allows the development of female hybrids, its activity being dependent on the temperature at which development occurs. Other similar genes have been located in distinct populations of species of this group. In short, only a few genes are needed for an effective post copulatory isolation barrier mediated through the non-viability of the hybrids.

As important as identifying an isolation gene is knowing its function. The Hmr gene, linked to the X chromosome and implicated in the viability of male hybrids between *D. melanogaster* and *D. simulans*, is a gene from the proto-oncogene family myb, that codes for a transcriptional regulator. Two variants of this gene function perfectly well in each separate species, but in the hybrid they do not function correctly, possibly due to the different genetic background of each species. Examination of the allele sequence of the two species shows that

change of direction substitutions are more abundant than synonymous substitutions, suggesting that this gene has been subject to intense natural selection.

The Dobzhansky-Muller model proposes that reproductive incompatibilities between species are caused by the interaction of the genes of the respective species. It has been demonstrated recently that *Lhr* has functionally diverged in *D. simulans* and will interact with *Hmr* which, in turn, has functionally diverged in *D. melanogaster* to cause the lethality of the male hybrids. *Lhr* is located in a heterochromatic region of the genome and its sequence has diverged between these two species in a manner consistent with the mechanisms of positive selection. An important unanswered question is whether the genes detected correspond to old genes that initiated the speciation favoring hybrid non-viability, or are modern genes that have appeared post-speciation by mutation, that are not shared by the different populations and that suppress the effect of the primitive non-viability genes. The *OdsH* (abbreviation of *Odysseus*) gene causes partial sterility in the hybrid between *Drosophila simulans* and a related species, *D. mauritiana*, which is only encountered on Mauritius, and is of recent origin. This gene shows monophyly in both species and also has been subject to natural selection. It is thought that it is a gene that intervenes in the initial stages of speciation, while other genes that differentiate the two species show polyphyly. *Odsh* originated by duplication in the genome of *Drosophila* and has evolved at very high rates in *D. mauritiana*, while its paralogue, *unc-4*, is nearly identical between the species of the group *melanogaster*. Seemingly, all these cases illustrate the manner in which speciation mechanisms originated in nature, therefore they are collectively known as "speciation genes", or possibly, gene sequences with a normal function within the populations of a species that diverge rapidly in response to positive selection thereby forming reproductive isolation barriers with other species. In general, all these genes have functions in the transcriptional regulation of other genes.

The *Nup96* gene is another example of the evolution of the genes implicated in post-copulatory isolation. It regulates the production of one of the approximately 30 proteins required to form a nuclear pore. In each of the *simulans* groups of *Drosophila* the protein from this gene interacts with the protein from another, as yet undiscovered, gene on the X chromosome in order to form a functioning pore. However, in a hybrid the pore that is formed is defective and causes sterility. The differences in the sequences of *Nup96* have been subject to adaptive selection, similar to the other examples of speciation genes described above.

Post-copulatory isolation can also arise between chromosomally differentiated populations due to chromosomal translocations and inversions. If, for example, a reciprocal translocation is fixed in a population, the hybrid produced between this population and one that does not carry the translocation will not have a complete meiosis. This will result in the production of unequal gametes containing unequal numbers of chromosomes with a reduced fertility. In certain cases, complete translocations exist that involve more than two chromosomes, so that the meiosis of the hybrids is irregular and their fertility is zero or nearly zero. Inversions can also give rise to abnormal gametes in heterozygous individuals but this effect has little importance compared to translocations. An example of chromosomal changes causing sterility in hybrids comes from the study of *Drosophila nasuta* and *D. albomicans* which are twin species from the Indo-Pacific region. There is no sexual isolation between them and the F1 hybrid is fertile. However, the F2 hybrids are relatively infertile and leave few descendants which have a skewed ratio of the sexes. The reason is that the X chromosome of *albomicans* is translocated and linked to an autosome which causes abnormal meiosis in hybrids. Robertsonian translocations are variations in the numbers of chromosomes that arise

from either: the fusion of two acrocentric chromosomes into a single chromosome with two arms, causing a reduction in the haploid number, or conversely; or the fission of one chromosome into two acrocentric chromosomes, in this case increasing the haploid number. The hybrids of two populations with differing numbers of chromosomes can experience a certain loss of fertility, and therefore a poor adaptation, because of irregular meiosis.

#### In plants

A large variety of mechanisms have been demonstrated to reinforce reproductive isolation between closely related plant species that either historically lived or currently live in sympatry. This phenomenon is driven by strong selection against hybrids, typically resulting from instances in which hybrids suffer reduced fitness. Such negative fitness consequences have been proposed to be the result of negative epistasis in hybrid genomes and can also result from the effects of hybrid sterility. In such cases, selection gives rise to population-specific isolating mechanisms to prevent either fertilization by interspecific gametes or the development of hybrid embryos.

Because many sexually reproducing species of plants are exposed to a variety of interspecific gametes, natural selection has given rise to a variety of mechanisms to prevent the production of hybrids. These mechanisms can act at different stages in the developmental process and are typically divided into two categories, pre-fertilization and post-fertilization, indicating at which point the barrier acts to prevent either zygote formation or development. In the case of angiosperms and other pollinated species, pre-fertilization mechanisms can be further subdivided into two more categories, pre-pollination and post-pollination, the difference between the two being whether or not a pollen tube is formed. (Typically when pollen encounters a receptive stigma, a series of changes occur which ultimately lead to the growth of a pollen tube down the style, allowing for the formation of the zygote.) Empirical investigation has demonstrated that these barriers act at many different developmental stages and species can have none, one, or many barriers to hybridization with interspecifics.

#### Examples of pre-fertilization mechanisms

A well-documented example of a pre-fertilization isolating mechanism comes from study of Louisiana iris species. These iris species were fertilized with interspecific and conspecific pollen loads and it was demonstrated by measure of hybrid progeny success that differences in pollen-tube growth between interspecific and conspecific pollen led to a lower fertilization rate by interspecific pollen. This demonstrates how a specific point in the reproductive process is manipulated by a particular isolating mechanism to prevent hybrids.

Another well-documented example of a pre-fertilization isolating mechanism in plants comes from study of the 2 wind-pollinated birch species. Study of these species led to the discovery that mixed conspecific and interspecific pollen loads still result in 98% conspecific fertilization rates, highlighting the effectiveness of such barriers. In this example, pollen tube incompatibility and slower generative mitosis have been implicated in the post-pollination isolation mechanism.

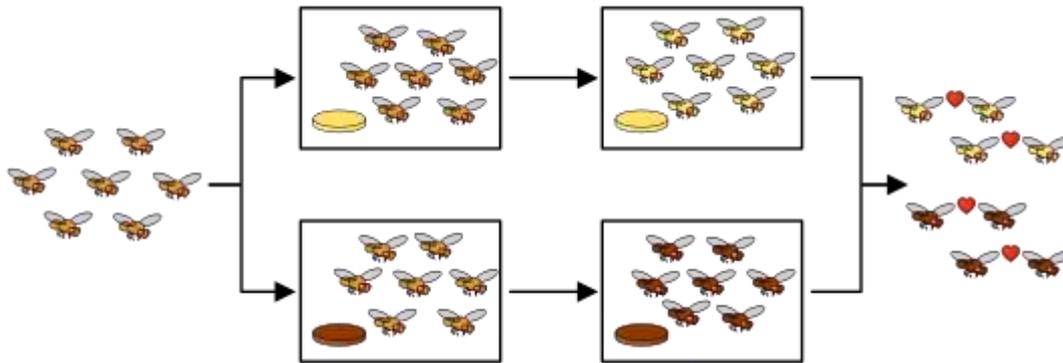
#### Examples of post-fertilization mechanisms

Crosses between diploid and tetraploid species of *Paspalum* provide evidence of a post-fertilization mechanism preventing hybrid formation when pollen from tetraploid species was used to fertilize a female of a diploid species. There were signs of fertilization and even endosperm formation but subsequently this endosperm collapsed. This demonstrates evidence of an early post-fertilization isolating mechanism, in which the hybrid early embryo is detected and selectively aborted. This process can also occur later during development in which developed, hybrid seeds are selectively aborted.

#### Selection

The sexual isolation between *Drosophila miranda* and *D. pseudoobscura*, for example, is more or less pronounced according to the geographic origin of the flies being studied. Flies

from regions where the distribution of the species is superimposed show a greater sexual isolation than exists between populations originating in distant regions.



Reproductive isolation can be caused by allopatric speciation. A population of *Drosophila* was divided into sub populations selected to adapt to different food types. After some generations the two sub populations were mixed again. Subsequent matings occurred between individuals belonging to the same adapted group. On the other hand, interspecific hybridization barriers can also arise as a result of the adaptive divergence that accompanies allopatric speciation. This mechanism has been experimentally proved by an experiment carried out by Diane Dodd on *D. pseudoobscura*. A single population of flies was divided into two, with one of the populations fed with starch-based food and the other with maltose-based food. This meant that each sub population was adapted to each food type over a number of generations. After the populations had diverged over many generations, the groups were again mixed; it was observed that the flies would mate only with others from their adapted population.

**Q.2.B. Answer Any Two of the following:**

**(10)**

**1. Discuss the role of mtDNA in tracing the origin of *Homo sapiens*.**

One important finding of mtDNA analyses is the corroboration of the “Recent African Origin” hypothesis of modern human origins, initially put forward on the basis of fossil evidence. Studies of mtDNA variation in worldwide populations have repeatedly found further evidence for this hypothesis, with the most recent common ancestor of human mtDNA located in Africa about 100,000–200,000 years ago. Moreover, direct analyses of mtDNA from fossils of Neandertals and their contemporaries, early modern humans from Europe, indicate no contribution of Neandertal mtDNA to modern humans.

Another insight gained from studies of mtDNA is a better understanding of the migrations that shaped human populations, such as the peopling of the New World, the colonization of the Pacific, the initial migration to New Guinea and Australia, and the settlement of Europe. However, mtDNA is only one locus, and only reflects the maternal history of a population; the history of a single locus may not accurately reflect the history of a population because of chance (drift) effects or because of selection acting on that locus. It has therefore become abundantly clear that studies of mtDNA variation need to be complemented with data on the male-specific Y-chromosome, and ideally with autosomal data as well. The use of mtDNA alone in studies of human evolution will probably decrease in the future, which indicates that studies of human variation are maturing beyond the single locus approach. However, the field has recently been confronted with several serious issues that need careful consideration. First, there is the increasing realization that nuclear inserts of mtDNA (numts) are more common than formerly thought. Analyses of the human genome sequence have revealed between 250

and more than 600 such inserts of differing length, with the largest fragment encompassing nearly the whole mitochondrial genome. Because mitochondrial inserts in the nuclear genome evolve at the nuclear rate, which is lower than that of mtDNA, known numts may have their uses as “molecular fossils”. However, a problem with numts is that they may not be detectable with standard methods and there are examples of numts that were mistakenly thought to represent authentic mtDNA sequences, leading to erroneous phylogenetic or medical conclusions precisely because of their slower rate of evolution. A second problem is the lack of quality control in published data. Reports of incorrect mtDNA sequences being published and submitted to databases are increasing in number, and even forensic databases have been accused of containing erroneous data. However, not all purported errors are truly errors, and the phylogenetic and forensic conclusions are rarely severely affected. Nevertheless, it is highly desirable that the data used in population genetic and evolutionary studies are of the highest quality, and statistical methods that aid in the detection of sequencing artefacts can be useful and should be used routinely. The rate of nonsynonymous mutations at several mitochondrial genes is higher than that of synonymous mutations within humans, but not between humans and chimps. This could be due to most mutations being slightly deleterious, so that they would contribute to polymorphism within populations, but would not become fixed and so would not contribute to differences between species.

However, the apparent excess of polymorphism within species could actually reflect an underestimate of the true divergence between species, due to the inability to detect recurrent mutations, which have occurred since the divergence of chimpanzees and humans.

It has also been suggested that the geographical variation seen in mtDNA haplogroups may reflect selection acting on specific lineages as humans spread from Africa to different climatic conditions. In particular, it has been claimed that haplogroups A, C, and D, which are characteristic of Siberians and Native Americans, have been strongly influenced by selection to uncouple the heat production and ATP production aspects of mitochondrial oxidative phosphorylation in order to produce more heat in colder climates. However, these provocative claims rest largely on statistical analyses of the distribution of nonsynonymous versus synonymous mutations in a phylogenetic tree of complete mtDNA sequences from the various haplogroups; detailed biochemical analyses are required to determine if there indeed has been a functional change in oxidative phosphorylation in individuals with these haplogroups. Because the A, C, and D haplogroups also occur in tropical environments in the Americas, where they have presumably been for at least 10,000 years, a further test of this hypothesis would be to see if these haplogroups have undergone further mutations in the Americas to more tightly couple heat and ATP generation. There also is evidence of regional violations of clock-like evolution of mtDNA in Africa. The overall consensus is that mainly purifying selection has been acting on human mtDNA; because the unit of inheritance is the whole genome, which does not undergo recombination, the noncoding control region is subject to the same selective forces as the coding DNA. This means that care must be taken when using mtDNA data to date phylogenetic events, as the underlying assumption of neutral clock-like evolution may not hold. However, the basic results of mtDNA studies, such as a recent initial migration out of Africa and subsequent world-wide dispersal of modern humans, are not affected by non-neutral evolution, and, furthermore, are supported by a wide array of other loci.

Even though mtDNA will probably be used less and less as the sole marker for elucidating human evolution and population history, it is still important for a wide range of questions. First of all, it is useful for unraveling socio-cultural effects that might have influenced human evolution, such as polygyny, the effects of matrilocality versus patrilocality or the social stratification induced by the caste system. Furthermore, because of the high copy number of

mtDNA versus the diploid autosomes and haploid Y-chromosome, mtDNA is crucial in studies of ancient DNA and in some applications of forensics. Depending on the age of the fossil sample, often only mtDNA will still be present, and therefore this is the only insight one can get into the genetic affinities of ancient populations

Finally, mtDNA is increasingly used in so-called personalized genetic histories. This is the use of genetic testing to investigate individual genealogies, including tracing the origins of immigrant/slave ancestors. The reality is that current mtDNA databases are not sufficiently detailed to allow such a high degree of geographic resolution, and even if they were, mtDNA types in general do not show sufficient geographic specificity to allow one to pinpoint such a specific origin. Furthermore, many customers inevitably confuse the place of origin of their maternal lineage with the place of origin of all of their biological ancestry, with potentially profound effects on how they view themselves and their identity. It must be kept in mind that mtDNA comprises only 0.0006% of the total human genome; as informative as mtDNA analyses have been and will continue to be for understanding human population history and evolution, when it comes to questions about personal genetic ancestry (as well as questions about human population history and evolution), it would be desirable to have some information on the remaining 99.9994% of the genome.

## **Q. 2 B.**

### **2. What advantages does vocalization confer over other modes of communication?**

Language is a unique hallmark of the human species. Although many species can communicate in limited ways about things that are physically present, only humans can construct a full narrative characterization of events occurring outside of the here and now. Humans are also unique in their ability to fashion tools such as arrow points, axes, traps, and clothing. By using language to control the social coordination of tool making, humans have produced a material society that has achieved domination over all the creatures of our world and often over Nature herself. The religions of the world have interpreted our unique linguistic endowment as a Special Gift bestowed directly by the Creator. Scientists have also been influenced by this view of language, often attributing the emergence of this remarkable species-specific ability to some single, pivotal salutatory event in human evolution. It seems quite likely that some aspect of language evolution played a major role in the recent creativity explosion. However, it would be a mistake to think that language could emerge suddenly in all its complex phonological and syntactic glory in the last 40,000 years without having been foreshadowed by major developments during the rest of our 6 million year history. In particular, we know that 300,000 years ago there was a major expansion of the parts of the vertebrae that carry nerves for the intercostal muscles (MacLarnon & Hewitt, 1999). The intercostals are the muscles that control the pulmonic pulsing that drives human phonation. The expansion of these pathways indicates that we were developing a reliance on vocal communication as far back as 300,000 years ago. But it is also apparent that the language we produced at that time was not structured or complex enough to serve as a support for the development of material culture.

Language relies on far more physiological, social, and neural systems than just the intercostal muscles. It depends on systems for cortical control of vocalization, changes in group structure and affective relations, growth in cognitive abilities, neural pathways for information integration, and mechanisms for the formation of social hierarchies. The hominid lineage has undergone a remarkable series of physiological adaptations involving skeletal modifications to support upright posture, development of an opposing thumb, changes in the birth process (Hockett & Ascher, 1964), loss of hair (Morgan, 1997), adaptation of the gastrointestinal tract, increased innervation of the intercostals muscles (MacLarnon & Hewitt, 1999), loss of

pronounced canine teeth, bending of the vocal tract, refinement of the facial musculature, freeing of the vocal folds, and sharpening of the chin. Each of these adaptations plays a role in supporting language. Beginning about 3 MYA (million years ago) there has been a gradual tripling of brain size (Holloway, 1995) which has brought massive changes in the interconnectedness of the frontal lobes, changes in the linkage of vocal production to motor and emotional areas, linkages of the visual areas to motor areas, and expansion of many older areas, including the cerebellum, basal ganglion, and thalamus. These various neurological developments have also provided a basis for a marked increase in humans' ability to actions through movement and sounds through vocalization. Alongside these changes in morphology and neurology, human society has undergone a parallel process of development involving the expansion of social groups, migrations first across Eurasia and then to the Americas, the refinement of warfare, the development of tools, and the emergence of language. This evolutionary analysis is designed to provide a basic account of the evolution of language in our species.

### **Q. 2 B)**

#### **3. What are the advantages of bipedalism?**

Bipedalism allowed hominids to free their arms completely, enabling them to make and use tools efficiently, stretch for fruit in trees and use their hands for social display and communication. They could also see further over the savannah grass – but this also could have been a disadvantage since predators could probably spot them more easily. Bipedal hominids could spend more time foraging and scavenging out in the open savannah because their bodies would be exposed to less sunlight standing upright. Bipedalism allowed hominids to free their arms, allowing the use of tools. Walking on two limbs was also more energy efficient than walking on four – giving early hominids more energy to reproduce and therefore more chance of producing offspring bearing this unique trait. But even with these advantages, these transitional hominids probably spent time in the trees as well. At least some *Australopithecus* species, including the one represented by “Little Foot” at Sterkfontein, which is as yet unnamed, were at least partly arboreal between 4-million and 3-million years ago, when there was some forest in the Cradle of Humankind environment. Similarly, further north in Africa, the *Australopithecus* species of Ethiopia and Tanzania between 3-million and 2-million years ago would have been able to climb trees better than modern humans, but were simultaneously adapting to more full-time upright walking. *Australopithecus afarensis*, which populated the Afar Depression in Ethiopia, would have lived in an environment typified by wetlands, woodland and forest. But the bipedal footprints of *Australopithecus afarensis* in Laetoli, Tanzania, are found in an area where the environment was probably drier and sparsely wooded 3.6-million years ago. “Little Foot”, which represents a species of *Australopithecus* more than 3.3-million years old, was most certainly not a knuckle-walker like some of the great apes. It probably could have walked and climbed effectively. “Little Foot” and other early australopithecines probably climbed trees to escape predators and maybe even to sleep in at night.

### **Q. 2 B )**

#### **4. Discuss Altruism w.r.t Human Evolution.**

Altruistic behaviours appear most obviously in kin relationships, such as in parenting, but may also be evident among wider social groups, such as in social insects. They allow an individual to increase the success of its genes by helping relatives that share those genes. Obligate altruism is the permanent loss of direct fitness (with potential for indirect fitness gain). For example, honey bee workers may forage for the colony. Facultative altruism is

temporary loss of direct fitness (with potential for indirect fitness gain followed by personal reproduction); for example, a Florida scrub jay helping at the nest, then gaining parental territory. Humans are an intensely social species, frequently performing costly behaviors that benefit others. Kinship, reciprocity, indirect reciprocity, punishment, and morality. Humans do not calculate the profits of altruism, but pay the costs or run the risks of it because they have human altruistic emotions.

Altruism in humans confirm to the "four propositions" offered on a preceding page? I think it does; at least this is a good working hypothesis. Human altruism does seem to be potentially reciprocal and profitable; some individuals do happen to lose by it, but all "expect" (are statistically likely to receive) profits that exceed their costs, although the expectation is usually not conscious, and although the profits are (and are expected to be) unequal. Altruists and recipients are environmentally determined, by position in groups or environmental situations; no persons are condemned by their genes to be altruists with no chance of being recipients. Altruism is therefore a net-gain lottery for humans as well as for other organisms. A man is a man and behaves as one because of his genes. However, no specific "genes for altruism" have been found in humans; "sociobiologists" are sometimes accused of postulating them, but rarely do. (However, behavior is a function of the whole organism, and many genes are known that change structures, physiologies, or endocrine systems in ways that change behaviors.)

Parent-offspring altruism is conspicuous and its importance unquestioned. Altruism is conspicuous also among human sibs and less-close kin, but it is not precisely proportional to kinship; it is greatly modified, either reinforced or cancelled, by individual likes and dislikes. Among young adults, among whom kin altruism should be most profitable genetically, it is overridden by attractions and reciprocal altruism between non-kin males and females. In general, altruism in humans seems correlated with congruity and compatibility more than with kinship, and this suggests that human altruism is responsive more than kin-related.

Altruistic behaviours do not fossilize, and evidence derived from observation of surviving hunter-gatherers and nonhuman primates is indirect and incomplete, so the answer must be largely inference and outright guessing. We can guess that among our remote prehuman ancestors altruistic behaviors may have been determined in some detail by specific genes and may have evolved by genetic variation and selection, but now, although our altruism still has a broad genetic base, the details of it and its evolution seem to be determined socially more than genetically. This has involved a shift to evolution at a new, social level. Variation and selection occur at the new level, but with new characteristics: the variation is amplified by something like inheritance of acquired characters, which greatly increases the rates and amounts of diversification that occur before selection; and, although selection is still primarily by elimination, it can be supplemented by intelligent choice.

Group selection does supplement individual selection in evolution of altruism in humans; this is illustrated by control of "cheating." "Cheating," or acceptance of altruism without reciprocation, does occur in human populations. Milder forms of it, including minor failures of responses or cooperation, are recognized and countered or tolerated by individuals. More serious forms are controlled by laws or other group actions. However, control is not perfect. Human populations usually carry loads of non altruists, and if the loads become too great, whole populations may be eliminated; this results in a kind of group selection which has presumably continually supplemented individual selection in evolution of altruism in humans, and may still operate. "Forced altruism," including outright slavery, also occurs in human beings, and does sometimes evolve into reciprocal altruism.

**Q.3 A Answer any 1****10M****1.**

Patient serial no.	On admission x	At 24 months Y	d=X-Y	d <sup>2</sup>
1	40	52	-12	144
2	41	43	-2	4
3	38	46	-8	64
4	41	52	-11	121
5	40	46	-6	36
6	37	38	-1	1
7	39	42	-3	9
8	37	41	-4	16
9	41	42	-1	1
10	35	38	-3	9
			$\sum d = -51$	$\sum d^2 = 405$

Null hypothesis: there is no significant difference between the two weights, ie  $\mu_x = \mu_y$

Alternative hypothesis  $\mu_x \neq \mu_y$  two tailed test

Mean d =  $51 / 10 = -5.1$

$$S^2 = \frac{1}{n-1} [\sum d^2 - (\sum d)^2 / n]$$

$$= \frac{1}{9} (405 - 2601 / 10)$$

$$= 16.1$$

Standard Error =  $S / \sqrt{n} = 1.29$

Student's t = Mean d / Standard error

$$= -5.1 / 1.29$$

$$= -3.95$$

$$|t| = 3.95$$

Given  $t_{tab} = 2.26 < t_{cal}$

Therefore null hypothesis is rejected that is there is significant difference in weights as an effect of drug.

2. Null hypothesis: there is no difference between Group A and B students hearing levels

i.e.  $\mu_A = \mu_B$

Alternative hypothesis  $\mu_A \neq \mu_B$  Two tailed test

$$t = \frac{\text{Mean XA} - \text{Mean XB}}{S \times \sqrt{\frac{n_A n_B}{n_A + n_B}}}$$

$$S = \sqrt{\frac{n_A S_A^2 + n_B S_B^2}{n_A + n_B - 2}}$$

$$= 7.04$$

$$t = \frac{10.5 - 20}{7.04 \times 2.63}$$

$$= -3.5$$

$$|t| = 3.5$$

Given t value = 2.055 < t cal

Therefore null hypothesis rejected that is there is significant difference between the hearing levels of group A and group B

**Q.3.B. Answer any 2**

**10M**

1.

No mutation	BRCA1 mutations	BRCA2 mutations
2.5	1.3	1.9
3.7	1.0	1.9
4.2	1.1	1.7
Mean = 3.4	Mean = 1.1	Mean = 1.8

Combined mean = 2.1

$$SS \text{ between} = 3(3.4 - 2.1)^2 + 3(1.1 - 2.1)^2 + 3(1.8 - 2.1)^2$$

$$= 8.34$$

$$MS \text{ between} = 8.34 / 2 = 4.17$$

$$SS \text{ within} = \sum(X1-\text{MeanX})^2 + \sum(X2-\text{MeanX})^2 + \sum(X3-\text{MeanX})^2$$

$$= 1.54 + 0.05 + 0.03 = 1.62$$

$$MS \text{ within} = 1.62 / 6 = 0.27$$

$$F = 4.17 / 0.27 = 15.44$$

$$\text{Given } F_{\text{tab}} = 5.14 < F_{\text{cal}}$$

Therefore the difference in mutations is significant between 3 groups.

### Q 3 B )

2. Given  $n=210$ ,  $\text{meanx}= 20$ ,  $s= 6$ ,  $\mu=19.2$

Null hypothesis :  $\mu=19.2$

Alternative hypothesis  $\mu \neq 19.2$

Test  $z = \text{mean X} - \mu / \text{Standard error}$

$$= 20 - 19.2 / 6/\sqrt{210}$$

$$= 0.8 / 0.41 = 1.95$$

Given  $z_{\text{tab}} = 1.96 > z_{\text{cal}}$  , Null hypothesis accepted,  $\mu=19.2$

### Q 3. B)

3. Observed frequency

	Pass	Fail	
Attended	29	6	35
Skipped	8	15	23
	37	21	58

Expected frequency

$35 \times 37 / 58 = 22.32$	$35 \times 21 / 58 = 12.67$	34.99
$23 \times 37 / 58 = 14.67$	$23 \times 21 / 58 = 8.32$	22.99
36.99	20.99	57.98

$$\Psi^2 = \sum(O-E)^2 / E$$

$$= (29-22.32)^2 / 22.32 + (6-12.67)^2 / 12.67 + (8-14.67)^2 / 14.67 + (15-8.32)^2 / 8.32$$

$$= 13.89$$

Given Chi square = 3.84 < Chi square cal

Null hypothesis is rejected that is there is association between attendance and passing among students.

### **Q. 3. B)**

#### **4. Uses and assumptions of parametric test**

Student's t test

Uses:

To test the significance of a single mean, when the population variance sigma is unknown

To test the significance of difference between two sample means the population variances being equal and unknown.

To test the significance of an observed sample correlation coefficient or difference between means of two samples.

#### **T-Test Assumptions**

The first assumption made regarding t-tests concerns the scale of measurement. The assumption for a t-test is that the scale of measurement applied to the data collected follows a continuous or ordinal scale, such as the scores for an IQ test.

The second assumption made is that of a simple random sample, that the data is collected from a representative, randomly selected portion of the total population.

The third assumption is the data, when plotted, results in a normal distribution, bell-shaped distribution curve.

The fourth assumption is a reasonably large sample size is used. A larger sample size means the distribution of results should approach a normal bell-shaped curve.

The final assumption is homogeneity of variance. Homogeneous, or equal, variance exists when the standard deviations of samples are approximately equal.

**Q. 4 A) Answer Any One of the following:**

**(10)**

**1. With suitable examples, Explain the terms homologs, paralog and orthologs found in Gene families of Eukaryotes, explain the significance of Orthologs.**

**Homolog**

- A gene related to a second gene by descent from a common ancestral DNA sequence. The term, homolog, may apply to the relationship between genes separated by the event of speciation (see ortholog) or to the relationship between genes separated by the event of genetic duplication .

**Ortholog**

- Orthologs are genes in different species that evolved from a common ancestral gene by speciation. Normally, orthologs retain the same function in the course of evolution. Identification of orthologs is critical for reliable prediction of gene function in newly sequenced genomes.

**Speciation**

- Speciation is the origin of a new species capable of making a living in a new way from the species from which it arose. As part of this process it has also acquired some barrier to genetic exchange with the parent species.

**Paralog**

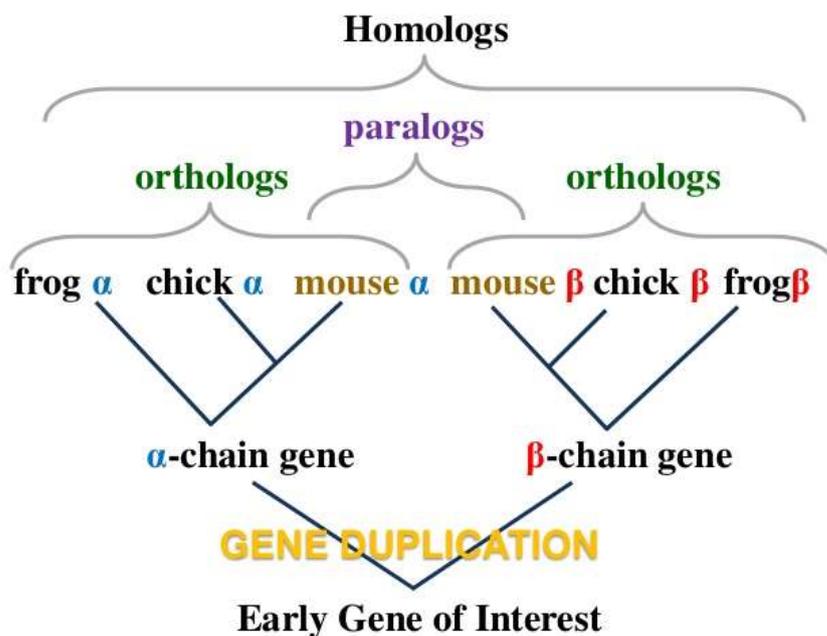
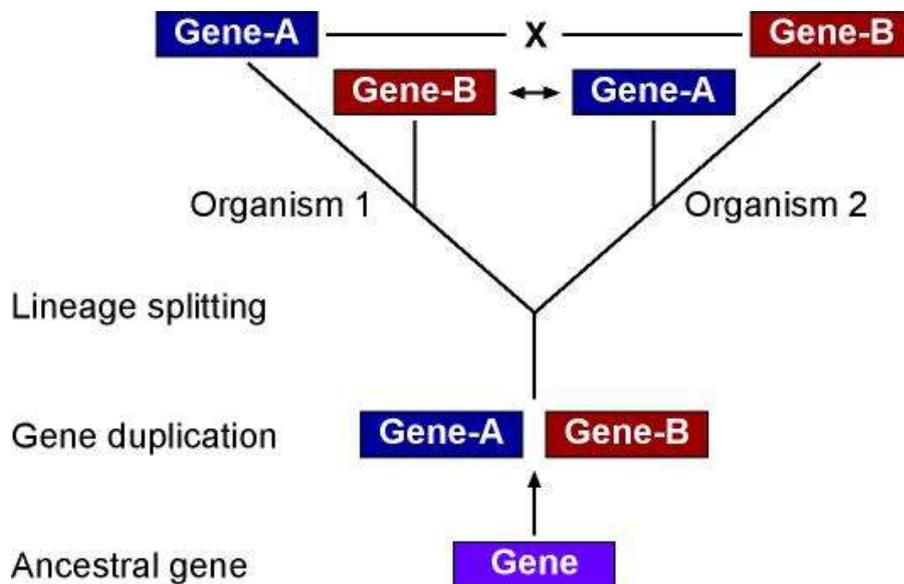
- Paralogs are genes related by duplication within a genome. Orthologs retain the same function in the course of evolution, whereas paralogs evolve new functions, even if these are related to the original one.

**Why does this study matter?**

In the absence of biochemical assays, the best possible inference for gene function is that it is shared by orthologs, and that gene duplications allow one copy to diverge to take on a new function or to be otherwise specialized (e.g., in timing or location of expression).

- The figure below illustrates how the commonly used method of reciprocal best-BLAST matching leads to incorrect assignment of gene identities (and their correlate, gene function). In this example (found for many real world examples), the evolutionary split between the two organisms has occurred after a gene duplication that generated paralogs named "Gene-A" and "Gene-B". Genes do not all evolve at the same rate and, in this example, we're imagining that it is Gene-B in organism 1 and Gene-A in organism 2 that happen to have the slower rates. That being the case, the reciprocal best matches are between Gene-B of organism 1 and Gene-A of organism 2, so these paralogs are erroneously inferred to be orthologous and assigned the same function. The other two genes are assigned no function at all, since the best match to Gene-A of organism 1 is Gene-A of organism 2, but this is not reciprocal, and similarly for Gene-B of organism 2.

- Only a complete phylogenetic reconstruction using accurate methods - such as is done in the PHRINGE pipeline - can reconstruct this and make guide the proper inference of orthology and functional assignment.



Q. 4 A.

## 2. Essay on Phylogenetic Trees and their Significance.

### Phylogenetic Trees and their Types:

A phylogenetic tree or evolutionary tree is a branching diagram or "tree" showing the inferred evolutionary relationships among various biological species or other entities—their phylogeny—based upon similarities and differences in their physical or genetic characteristics. The taxa joined together in the tree are implied to have descended from a common ancestor. Phylogenetic trees are central to the field of phylogenetics.

In a rooted phylogenetic tree, each node with descendants represents the inferred most recent common ancestor of the descendants, and the edge lengths in some trees may be interpreted as time estimates. Each node is called a taxonomic unit. Internal nodes are generally called hypothetical taxonomic units, as they cannot be directly observed. Trees are useful in fields of biology such as bioinformatics, systematics, and phylogenetic comparative methods.

Unrooted trees illustrate only the relatedness of the leaf nodes and do not require the ancestral root to be known or inferred.

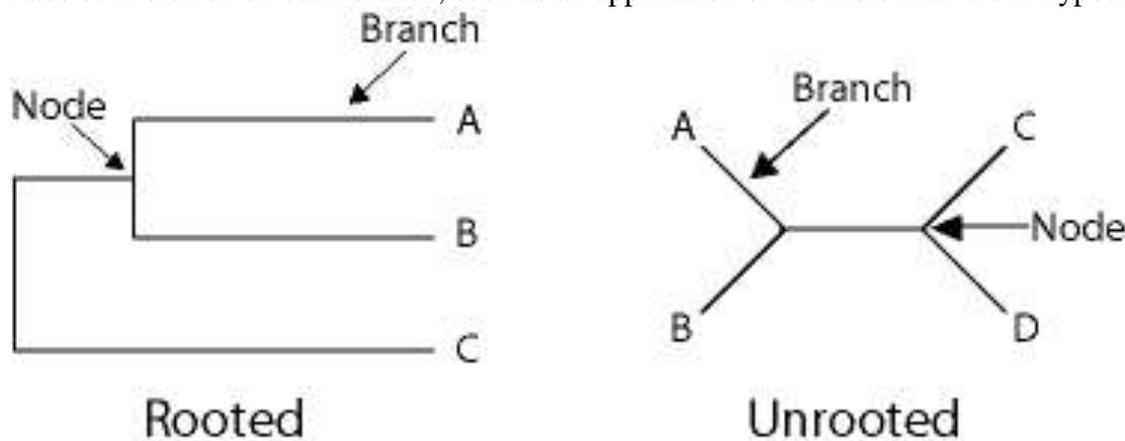
### Rooted Tree:

A rooted phylogenetic tree (see two graphics at top) is a directed tree with a unique node — the root — corresponding to the (usually imputed) most recent common ancestor of all the entities at the leaves of the tree. The root node does not have a parent node, but serves as the parent of all other nodes in the tree. The root is therefore a node of degree 2 while other internal nodes have a minimum degree of 3 (where "degree" here refers to the total number of incoming and outgoing edges).

The most common method for rooting trees is the use of an uncontroversial outgroup—close enough to allow inference from trait data or molecular sequencing, but far enough to be a clear outgroup. **Eg/Diagram**

### Unrooted tree:

An unrooted phylogenetic tree for myosin, a super family of proteins. Unrooted trees illustrate the relatedness of the leaf nodes without making assumptions about ancestry. They do not require the ancestral root to be known or inferred. Unrooted trees can always be generated from rooted ones by simply omitting the root. By contrast, inferring the root of an unrooted tree requires some means of identifying ancestry. This is normally done by including an outgroup in the input data so that the root is necessarily between the outgroup and the rest of the taxa in the tree, or by introducing additional assumptions about the relative rates of evolution on each branch, such as an application of the molecular clock hypothesis.



### Importance of Phylogenetic Analysis:

#### Phylogenetic Analysis can be useful in following fields:

**Classification:** Phylogenetics based on sequence data provides us with more accurate descriptions of patterns of relatedness than was available before the advent of molecular sequencing. Phylogenetics now informs the Linnaean classification of new species.

**Forensics:** Phylogenetics is used to assess DNA evidence presented in court cases to inform situations, e.g. where someone has committed a crime, when food is contaminated, or where the father of a child is unknown.

**Identifying the origin of pathogens:** Molecular sequencing technologies and phylogenetic approaches can be used to learn more about a new pathogen outbreak. This includes finding out about which species the pathogen is related to and subsequently the likely source of transmission. This can lead to new recommendations for public health policy.

**Conservation:** Phylogenetics can help to inform conservation policy when conservation biologists have to make tough decisions about which species they try to prevent from becoming extinct.

**Bioinformatics and computing:** Many of the algorithms developed for phylogenetics have been used to develop software in other fields.

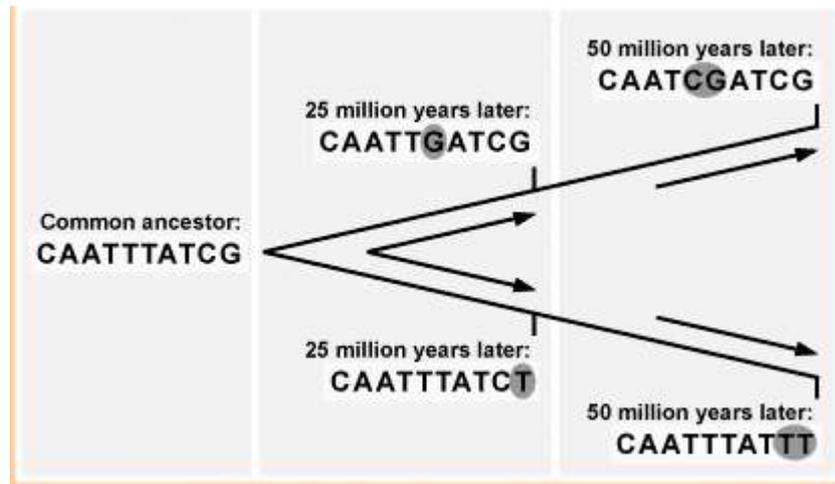
**Q. 4 B. Explain Any Two of the following: (10)**

**1. Evolutionary Clocks and their importance.**

**Evolutionary Clocks:**

The **Evolutionary Clock** is a technique that uses the [mutation rate](#) of [biomolecules](#) to [deduce the time](#) in [prehistory](#) when two or more [life forms diverged](#). The biomolecular data used for such calculations are usually nucleotide sequences for DNA or amino acid sequences for proteins. The benchmarks for determining the mutation rate are often fossil or archaeological dates. The molecular clock was first tested in 1962 on the hemoglobin protein variants of various animals, and is commonly used in molecular evolution to estimate times of speciation or radiation. It is sometimes called a **gene clock** or an **evolutionary clock**. The notion of the existence of a so-called "molecular clock" was first attributed to Émile Zuckerkandl and Linus Pauling who, in 1962, noticed that the number of amino acid differences in hemoglobin between different lineages changes roughly linearly with time, as estimated from fossil evidence. They generalized this observation to assert that the rate of evolutionary change of any specified protein was approximately constant over time and over different lineages (based on the **molecular clock hypothesis (MCH)**). The molecular clock technique is an important tool in molecular systematics, the use of molecular genetics information to determine the correct scientific classification of organisms or to study variation in selective forces. Knowledge of approximately constant rate of molecular evolution in particular sets of lineages also facilitates establishing the dates of phylogenetic events, including those not documented by fossils, such as the divergence of living taxa and the formation of

the phylogenetic tree. In these cases—especially over long stretches of time—the limitations of MCH (above) must be considered; such estimates may be off by 50% or more.



## 2. Significance of Six Frame Translation with suitable example.

In molecular genetics, an **open reading frame (ORF)** is the part of a reading frame that has the ability to be translated. An ORF is a continuous stretch of codons that contain a start codon (usually AUG) and a stop codon (usually UAA, UAG or UGA). An ATG codon within the ORF (not necessarily the first) may indicate where translation starts. The transcription termination site is located after the ORF, beyond the translation stop codon. If transcription were to cease before the stop codon, an incomplete protein would be made during translation.<sup>[2]</sup> In eukaryotic genes with multiple exons, ORFs span intron/exon regions, which may be spliced together after transcription of the ORF to yield the final mRNA for protein translation.

1. **ATG** CAA TGG GGA AAT GTT ACC AGG TCC GAA CTT ATT GAG GTA AGA CAG ATT **TAA**
2. A TGC AAT GGG GAA **ATG** TTA CCA GGT CCG AAC TTA TTG AGG **TAA** GAC AGA TTT AA
3. AT GCA **ATG** GGG AAA TGT TAC CAG GTC CGA ACT TAT **TGA** GGT AAG ACA GAT TTA A

Since DNA is interpreted in groups of three nucleotides (codons), a DNA strand has three distinct reading frames. The double helix of a DNA molecule has two anti-parallel strands; with the two strands having three reading frames each, there are six possible frame translations. One needs to consider *six* reading frames when considering the potential of DNA to encode protein (three frames for each strand). But only one strand is transcribed into RNA — the so-called *coding strand*. It would therefore seem to me that there are actually only *three* reading frames to consider. Explain with suitable example. Of +1,+2,+3 and -1,-2,-3 frames.

Reading  
frame

```

+3      L V R T
+2      T C S Y
+1      N L F V
5' -AACTTGTTTCGTACA-3'
3' -TTGAACAAGCATGT-5'
-1      K N T C
-2      S T R V
-3      V Q E Y

```

		Second base						
		U	C	A	G			
U	UUU	Phenylalanine <b>F</b>	UCU	Serine <b>S</b>	UAU	Tyrosine <b>Y</b>	UGU	Cysteine <b>C</b>
	UUC		UCC		UAC		UGC	
	UUA	Leucine <b>L</b>	UCA		UAA	Stop codon	UGA	Stop codon
C	CUU	Leucine <b>L</b>	CCU	Proline <b>P</b>	CAU	Histidine <b>H</b>	CGU	Arginine <b>R</b>
	CUC		CCC		CAC		CGC	
	CUA		CCA		CAA	Glutamine <b>Q</b>	CGA	
A	AUU	Isoleucine <b>I</b>	ACU	Threonine <b>T</b>	AAU	Asparagine <b>N</b>	AGU	Serine <b>S</b>
	AUC		ACC		AAC		AGC	
	AUA		ACA		AAA	Lysine <b>K</b>	AGA	Arginine <b>R</b>
G	GUU	Valine <b>V</b>	GCU	Alanine <b>A</b>	GAU	Aspartic acid <b>D</b>	GGU	Glycine <b>G</b>
	GUC		GCC		GAC		GGC	
	GUA		GCA		GAA	Glutamic acid <b>E</b>	GGA	
G	GUG		GCG		GAG		GGG	
	UUG		UCG		UAG		UGG	Tryptophan <b>W</b>
	UUG		UCG		UAG		UGG	

### Biological Significance:

One common use of open reading frames (ORFs) is as one piece of evidence to assist in gene prediction. Long ORFs are often used, along with other evidence, to initially identify candidate protein-coding regions or functional RNA-coding regions in a DNA sequence.<sup>[3]</sup> The presence of an ORF does not necessarily mean that the region is always translated. For example, in a randomly generated DNA sequence with an equal percentage of each nucleotide, a stop-codon would be expected once every 21 codons. A simple gene prediction algorithm for prokaryotes might look for a start codon followed by an open reading frame that is long enough to encode a typical protein, where the codon usage of that region matches the frequency characteristic for the given organism's coding regions. By itself even a long open reading frame is not conclusive evidence for the presence of a gene. On the other hand, it has been proven that some short ORFs (sORFs) that lack the classical hallmarks of protein-coding genes (both from ncRNAs and mRNAs) can produce functional peptides. 5'NTR of about 50% of mammal mRNAs are known to contain one or several sORFs. 64–75% of experimentally found translation initiation sites of sORFs are conserved in the genomes of human and mouse and may indicate that these elements have function. However, sORFs can often be found only in the minor forms of mRNAs and avoid the selection; the high conservatism of initiation sites may be connected with their location inside promoters of the relevant genes. Such kind of situation is characteristic of SLAMF1 gene, for example.

### 3. Types of Gene Annotation and their significance.

**DNA annotation** or **genome annotation** is the process of identifying the locations of genes and all of the coding regions in a genome and determining what those genes do. An annotation (irrespective of the context) is a note added by way of explanation or commentary. Once a genome is sequenced, it needs to be annotated to make sense of it. For DNA annotation, a previously unknown sequence representation of genetic material is enriched with information relating genomic position to intron-exon boundaries, regulatory sequences, repeats, gene names and protein products. This annotation is stored in genomic databases such as Mouse Genome Informatics, FlyBase, and WormBase. Educational materials on some aspects of biological annotation from the 2006 Gene Ontology annotation camp and similar events are available at the Gene Ontology website. The National Center for Biomedical Ontology ([www.bioontology.org](http://www.bioontology.org)) develops tools for automated annotation of database records based on the textual descriptions of those records.

As a general method, dcGO has an automated procedure for statistically inferring associations between ontology terms and protein domains or combinations of domains from the existing gene/protein-level annotations.

Genome annotation consists of three main steps: identifying portions of the genome that do not code for proteins

1. Identifying elements on the genome, a process called gene prediction, and
2. attaching biological information to these elements.

Automatic annotation tools try to perform all this by computer analysis, as opposed to manual annotation (a.k.a. curation) which involves human expertise. Ideally, these approaches co-exist and complement each other in the same annotation pipeline. The simplest way to perform gene annotation relies on homology based search tools, like BLAST, to search for homologous genes in specific databases, the resulting information is then used to annotate genes and genomes. However, nowadays more and more additional information is added to the annotation platform. The additional information allows manual annotators to deconvolute discrepancies between genes that are given the same annotation. Some databases use genome context information, similarity scores, experimental data, and integrations of other resources to provide genome annotations through their Subsystems approach. Other databases (e.g. Ensembl) rely on both curated data sources as well as a range of different software tools in their automated genome annotation pipeline.

*Structural annotation* consists of the identification of genomic elements.

- ORFs and their localization
- gene structure
- coding regions
- location of regulatory motifs

*Functional annotation* consists of attaching biological information to genomic elements.

- biochemical function
- biological function
- involved regulation and interactions
- expression

These steps may involve both biological experiments and *insilico* analysis. Proteogenomics based approaches utilize information from expressed proteins, often derived from mass spectrometry, to improve genomics annotations. A variety of software tools have been developed to permit scientists to view and share genome annotations. Genome annotation remains a major challenge for scientists investigating the human genome, now that the genome sequences of more than a thousand human individuals and several model organisms are largely complete. Identifying the locations of genes and other genetic control elements is often described as defining the biological "parts list" for the assembly and normal operation of an organism. Scientists are still at an early stage in the process of delineating this parts list and in understanding how all the parts "fit together". Genome annotation is an active area of investigation and involves a number of different organizations in the life science community which publish the results of their efforts in publicly available biological databases accessible via the web and other electronic means. Here is an alphabetical listing of on-going projects relevant to genome annotation:

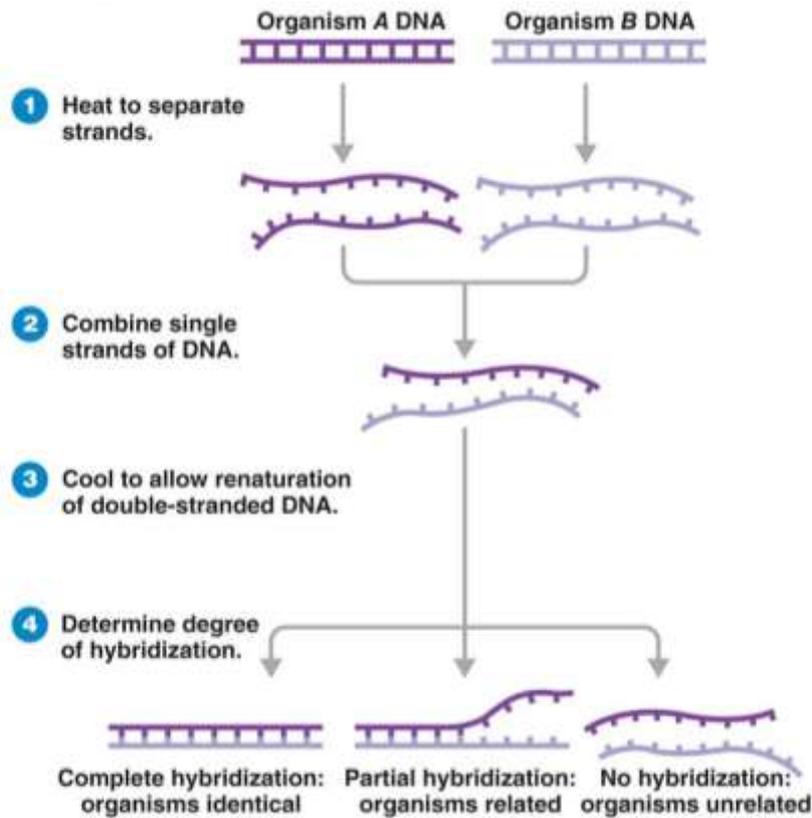
- Encyclopedia of DNA elements (ENCODE)
- Entrez Gene
- Ensembl
- GENCODE
- Gene Ontology Consortium
- GeneRIF
- RefSeq
- Uniprot
- Vertebrate and Genome Annotation Project (Vega)

#### 4. DNA hybridization and its significance in evolutionary studies.

**DNA–DNA hybridization** generally refers to a molecular biology technique that measures the degree of genetic similarity between pools of DNA sequences. It is usually used to determine the genetic distance between two organisms. This has been used extensively in phylogeny and taxonomy.

The DNA of one organism is labelled, then mixed with the unlabelled DNA to be compared against. The mixture is incubated to allow DNA strands to dissociate and then cooled to form renewed hybrid double-stranded DNA. Hybridized sequences with a high degree of similarity will bind more firmly, and require more energy to separate them: i.e. they separate when heated at a higher temperature than dissimilar sequences, a process known as "DNA melting". To assess the melting profile of the hybridized DNA, the double-stranded DNA is bound to a column and the mixture is heated in small steps. At each step, the column is washed; sequences that melt become single-stranded and wash off the column. The temperatures at which labelled DNA comes off the column reflects the amount of similarity between sequences (and the self-hybridization sample serves as a control). These results are combined to determine the degree of genetic similarity between organisms.

The modern approach is to carry out DNA–DNA hybridization *in silico* using completely or partially sequenced genomes.<sup>[9]</sup> The GGDC developed at DSMZ is the most accurate known tool for calculating DDH-analogous values.<sup>[9]</sup> Among other algorithmic improvements, it solves the problem with paralogous sequences by carefully filtering them from the matches between the two genome sequences.



**Q.5. Write short notes on any four of the following:**

**(20)**

**1.Social Evolution:**

Social evolution is actually the result of ‘group selection’, meaning the competition between groups organized according to different rules, that are selected on the basis of their functional adaptation.

a social process in some way analogous to the process of biological evolution. Different thinkers have had different aspects of biological evolution in mind (and sometimes, different conceptions of the nature of the biological process). In its most minimal sense, social and cultural evolution can just be thought of as social and cultural change. Social evolution is a process of directional social change, and evolutionary theories attempt to describe and explain this process. Theories of social evolution go back to the second half of the nineteenth century to Spencer, Morgan, Tylor, and Marx and Engels. After a lapse, evolutionary theorizing revived in the 1930s and 1940s with the work of Childe, White, and Steward, and continued into the 1960s and 1970s with the work of Sahlins, Service, Carneiro, Lenski, and Harris. Important typologies of stages of evolutionary development have been developed by most of these thinkers. Although there is far from complete consensus regarding the most important dimensions of social evolution, virtually everyone recognizes the Neolithic Revolution and the rise of civilization and the state as two extremely important evolutionary transformations.

**2.Punctuated Rate of Speciation:**

The theory of punctuated equilibrium argues that the mode of speciation is the result of reproductive isolation at the periphery of a species’ range, the emphasis being on cladogenesis as opposed to anagenesis (see Eldredge & Gould, 1972; Gould & Eldredge, 1977; Stanley, 1978, 1979, 1996; Eldredge, 1989; Gould, 2002). Cladogenesis is the splitting

of a single species into two reproductively isolated or genetically distinct lineages so that species remain relatively unchanged for long periods of time, occasionally interrupted by rapid or short bursts of evolutionary change resulting in speciation. The isolation of a marginalized population results in a rapid rate of speciation, which may be accompanied by the new daughter species taking over the parent species' territory. If this does occur, it is at this stage that we find the new species within the paleontological record. The daughter species is of course much more likely to be competitively inferior to the parent species and so to become extinct; but very occasionally it may outcompete or coexist with the parent species and become successful and abundant enough to become visible to us in the fossil record, having found its own niche, distinct from that of its parent species. This tempo and mode of evolution best fits the model of evolution espoused by those who support an "Out of Africa" origin for the hominins.

### 3. Null hypothesis and alternative hypothesis

**Null Hypothesis**—The statistical hypothesis that is set up for testing a hypothesis is known as null hypothesis. It is set up to decide whether to accept or reject the null hypothesis. It asserts that there is no difference between the sample statistic and population parameter. It is denoted by  $H_0$ .

Example of any z test or t test sum can be given

**Alternative hypothesis**—the negation of null hypothesis is called 'alternative hypothesis. When Null hypothesis is rejected, automatically alternative hypothesis is accepted. It is denoted by  $H_1$

e.g. we want to know the average height of college students is 165cm Null hypothesis is

$H_0: \mu=165$  and alternative hypothesis  $H_1: \mu \neq 165$

### 4. Comparison between parametric and non-parametric tests

BASIS FOR COMPARISON	PARAMETRIC TEST	NONPARAMETRIC TEST
Meaning	A statistical test, in which specific assumptions are made about the population parameter is known as parametric test.	A statistical test used in the case of non-metric independent variables, is called non-parametric test.

BASIS FOR COMPARISON	PARAMETRIC TEST	NONPARAMETRIC TEST
Basis of test statistic	Distribution	Arbitrary
Measurement level	Interval or ratio	Nominal or ordinal
Measure of central tendency	Mean	Median
Information about population	Completely known	Unavailable
Applicability	Variables	Variables and Attributes
Correlation test	Pearson	Spearman

### 5.Limitations of Phylogenetic Analysis:

**The biggest problem or limitations of a Phylogenetic tree building is that there are many different methods that construct trees.** The trees are often highly correlated. But sometimes they can have extremely different results and interpretations, because the methods answer very different questions. So like any analysis tool, they're quite fine if you use them properly (select the right tool/test, understand the assumptions, and consider the accuracy/error/variation). So the most consistent limitations is that a tree can't interpret every question you have. On the flip side, there is theoretically a specific tree for every question you have.

**The most common phylogenetic tree methods based on genetics are *not* genealogies** (they are not built for ancestry and pedigrees), even though they usually interpreted that way. They are similarities or clustering algorithms, which are useful for a snapshot of the current state in time. There are other methods for estimating genealogies that *do* go backwards, but they're much less common and more computationally intensive e.g. coalescent theory. The extra computation occurs because migration, recombination, mating, population size, and selection *can* be considered and built into the modeling.

**Rooting the tree is useful but biased and/or arbitrary.**

**The lengths can be misleading and there are a lot of ways to calculate them.**

**Some form of “variance”/error should be displayed (e.g. bootstrap values). They’re often not shown.**

**You can often construct very different but valid trees based on the same data.**

Depending on how they are displayed, **they can be easily misinterpreted**. This goes back to the main point about them being genealogies. The most common way to do this is to use the same tree, but change the linear order of the ends or nodes to represent a desired conclusion.

**Most trees cannot show horizontal transfer or recombination.** They rely on a binary tree structure.

It may be easy to assume that more closely related organisms look more alike, and while this is often the case, it is not always true. If two closely related lineages evolved under significantly varied surroundings or after the evolution of a major new adaptation, it is possible for the two groups to appear more different than other groups that are not as closely related. For example, the phylogenetic tree in Figure 1 shows that lizards and rabbits both have amniotic eggs, whereas frogs do not; yet lizards and frogs appear more similar than lizard and rabbits. Another aspect of phylogenetic trees is that, unless otherwise indicated, the branches do not account for length of time, only the evolutionary order. In other words, the length of a branch does not typically mean more time passed, nor does a short branch mean less time passed— unless specified on the diagram. For example, in Figure 1, the tree does not indicate how much time passed between the evolution of amniotic eggs and hair. What the tree does show is the order in which things took place. Again using Figure 1, the tree shows that the oldest trait is the vertebral column, followed by hinged jaws, and so forth. Remember that any phylogenetic tree is a part of the greater whole, and like a real tree, it does not grow in only one direction after a new branch develops. So, for the organisms in Figure 1, just because a vertebral column evolved does not mean that invertebrate evolution ceased, it only means that a new branch formed. Also, groups that are not closely related, but evolve under similar conditions, may appear more phenotypically similar to each other than to a close relative—lizards and rabbits.

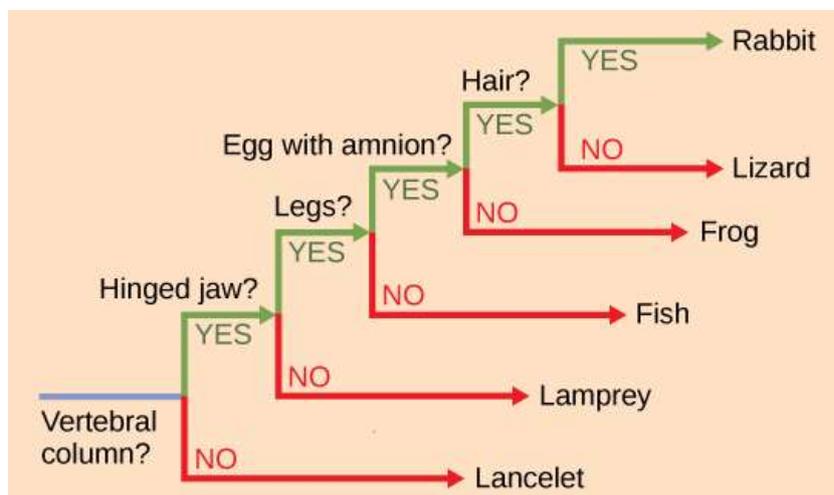


Figure 1. This ladder-like phylogenetic tree of vertebrates is rooted by an organism that lacked a vertebral column. At each branch point, organisms with different characters are placed in different groups based on the characteristics they share.

## 6. Nucleic acid sequence comparison and their evolutionary significance.

A nucleic acid sequence is a succession of letters that indicate the order of nucleotides forming alleles within a DNA (using GACT) or RNA (GACU) molecule. By convention, sequences are usually presented from the 5' end to the 3' end. For DNA, the sense strand is used. Because nucleic acids are normally linear (unbranched) polymers, specifying the sequence is equivalent to defining the covalent structure of the entire molecule. For this reason, the nucleic acid sequence is also termed the primary structure. By comparing two nucleic acid sequences, we can study the similarities and modifications between the sequences during the process of evolution. This helps in tracing lineages, common ancestry, mutations and events of speciation between species at Molecular levels. There are many software tools available to compare molecular sequences. (Eg. BLAST or any suitable software studied by student.)

Sequence analysis in molecular biology includes:

1. The comparison of sequences in order to find similarity, often to infer if they are related (homologous)
2. Identification of intrinsic features of the sequence such as active sites, post translational modification sites, gene-structures, reading frames, distributions of introns and exons and regulatory elements.
3. Identification of sequence differences and variations such as point mutations and single nucleotide polymorphism (SNP) in order to get the genetic marker.
4. Revealing the evolution and genetic diversity of sequences and organisms.
5. Identification of molecular structure from sequence alone.

In evolutionary biology, **conserved sequences** are identical or similar sequences in nucleic acids (DNA and RNA) or proteins across species (orthologous sequences), or within a genome (paralogous sequences), or between donor and receptor taxa (xenologous sequences). Conservation indicates that a sequence has been maintained by natural selection.

In coding sequences, the nucleic acid and amino acid sequence may be conserved to different extents, as the degeneracy of the genetic code means that synonymous mutations in a coding sequence do not affect the amino acid sequence of its protein product. Amino acid sequences can be conserved to maintain the structure or function of a protein or domain. **Significance in studying** Phylogenetics and taxonomy, Medical Research, Structural and Functional annotations.

Partial amino acid sequence comparison among  
glycosyltransferases in the ABO gene family  
(2001)

		* *	
Human A101	FTYERRPQSQAYIPKDEGDFYYLGGFFGG		272
Human B101	FTYERRPQSQAYIPKDEGDFYYMGAFFGG		272
Human O03	FTYERRPQSQAYIPKDEGDFYYLGRFFGG		272
Human cis-AB	FTYERRPQSQAYIPKDEGDFYYLGAFFGG		272
Mouse AB	FTYERRPQSQAYIPWDRGDFYYGGAFFGG		251
Pig A	FTYERRPLSQAYIPRDEGDFYYAGGFFGG		282
Dog Forssman	FPYERRHISTAFVAENEGDFYYCGAVFFGG		267
Mouse Galt	FTYERRELSAAYIPFGEGDFYYHAAIFFGG		312
Bovine Galt	FTYERRKESAAYIPFGEGDFYYHAAIFFGG		286

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