UNIT 13

HUMAN EVOLUTION

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13.1 INTRODUCTION

The process by which humans developed from extinct primates to the present day is called as Human evolution. Humans are *Homo sapiens*, who developed about 315000 years ago. This process entails the gradual emergence of features like human bipedalism and language. Human evolution was not linear but rather web-like. Humans were preceded by other hominins such as *Ardipithecus, Australopithecus*, and other *Homo* species.

Objectives

After studying this Unit, you would be able to:

- explain the history of evolution of *Homo sapiens*;
- understand the tree of life;
- describe the correlation between the genomics and humanness;

- comprehend the trends in general morphology during human evolution; and
- explain the unique phenotypes of modern humans.

13.2 HISTORY OF EVOLUTION OF *HOMO SAPIENS*

Neanderthal remnants were discovered in a limestone quarry in 1856 as the first fossil. Raymond Dart described *Australopithecus africanus* in 1925. The Taung Child, an australopithecine newborn was unearthed in a cave. The child's remains consisted of a very well-preserved small skull and a brain endocast. The brain was spherical, unlike those of chimps and gorillas, and looked more like a modern human brain. The species also had short canine teeth, and the position of the foramen magnum (the opening in the skull through which the spine enters) indicated bipedalism. Dart was sure that the Taung Child was a bipedal human ancestor, a transitional form between apes and humans.

The evolution process involves a series of changes that cause the species to either adapt to the environment or become extinct. Evolution is the result of changes in the genetic material. It does not change a single organism, but the entire group of organisms belonging to the same species *Homo sapiens*. The most prominent species on earth is the result of over 7 million years of evolution. Man belongs to the family Hominidae of the order Primates. The first-ever ancestors of humans are believed to have originated in Africa, eventually migrating to Europe, Asia and the rest of the world.

In the African continent around 200,000 years ago genus *Homo* was originated. Around 135,000 years ago *Homo erectus*, an extinct species lived on the Earth. The *Homo erectus* is the probably most the recent common ancestor of the human species. The only surviving species of the genus *Homo* is *Homo sapiens.*

Ardipithecus were one of the earliest ancestors to have been discovered, with fossils dating back more than 4.4 million years old.

***Homo sapiens* originated through several stages as given below:**

- ***Dryopethicus -*** *Dryopithecus* is considered to be the earliest known ancestor of man. They were found in some parts of Africa, Asia, and Europe. It has been known that the evolution of man began from him. Dryopethicus was being followed by *Australopithecus.*
- ***Australopithecus -*** *Australopithecus* could walk upright and they were 1.2 meters tall. They have inhabited the mainland of Africa. They used to have human-like teeth and had large jaws.
- ***Homo habilis -*** *Homo habilis* used to have a height of around 5 feet and they could even make use of tools. It has been believed that they were able to speak.

- ***Homo erectus -*** It has been considered that *Homo erectus* was more evolved. They used to have larger brains and they were also upright. They were the ones who invented the fire and were carnivorous. They also had a prominent speech.
- ***Homo sapiens -*** *Homo sapiens* are known as modern men. Home sapiens had developed the power of thinking and using the tools. They were omnivorous and were able to produce art. Their brain size was reduced to 1300 cc.

13.2.1 The Human Evolution

The process of human evolution can be studied from the remains of the earliest ancestors and races. Several skulls and skeletal parts of the hominids have been found in the Mid-Pleistocene or earlier in Transvaal, South Africa in 1925 and Olduvai Gorge Tanganyika in 1959. They possess characteristics features as mentioned in Table 13.1.

Table 13.1: Characteristics features of hominids.

Sr. No.	Organ/body part	Characteristics feature
1.	Skulls	Smaller in size with rounded rear part
2.	The volume of the brain	Between 600-700 c.c
3.	The forehead	Higher than in apes
4.	Face	Protruding
5.	Brow ridges	Prominent
6.	Occipital condyles	Ventrally placed
7.	Jaws	Large with small incisors
8.	Teeth	Large canines and large cheek teeth

Java Man - *Pithecanthropus erectus*

In the Mid-Pleistocene of Solo River near Trimil, Java the fragment remains of *Pithecanthropus erectus* were found from 1891 until 1945. *Pithecanthropus erectus* had ability of speech which was indicated by the imprint of the brain. They possess characteristics features as mentioned in Table 13.2.

Table 13.2: Characteristics features of *Pithecanthropus erectus*.

Sr. No	Organ/body part	Characteristics feature
1	Skull	A flattish-topped and projected behind.
2	The brow ridges	Solid above the orbits.
3	The brain volume	Between 775-900 c.c.
4	The jaws	Protruding
5	Teeth	Arranged in a curve.

Peking Man - *Pithecanthropus (Sinanthropus) pekinensis*

From the Mid-Pleistocene caves of Choukoutien (South-west of Peking), China, the parts of the skeletal structure and the remains of skulls of *Pithecanthropus (Sinanthropus) pekinensis* were found up to 1943. Signs of equipment of quartz and other rocks were discovered. The fireplace indicated the use of fire. Peking Man might have ability of speech. They possess characteristics features as mentioned in Table 13.3.

Table 13.3: Characteristics features of *Pithecanthropus (Sinanthropus) pekinensis*.

Sr. No	Organ/body part	Characteristics feature
1	The skull	Small and low-vaulted
2	The brain volume	850-1300 c.c
3	The brow ridges	Stout.

Transitional Man - *Homo habilis*

In the Pleistocene bed of East Africa the remnants of the transitional man were found. They could make the simple stone tools. *Homo habilis* is said to be of intermediate stage between the *Australopithecus* and *Pithecanthropus erectus.* The brain's average capacity was 680 c.c.

Heidelberg Man - *Homo heidelbergensis*

In 1907 in Germany near Heidelberg at Mauer one lower jaw of *Homo heidelbergensis* was discovered in a sandpit. Those remains were of the Mid-Pleistocene period. They possess characteristics features as mentioned in Table 13.4.

Table 13.4: Characteristics features of *Homo heidelbergensis*.

Sr. No.	Organ/body part	Characteristics feature
1	Chin.	Absent
2	Teeth	Stout, the canines not enlarged.
3	Jaw	Massive with very wide ascending ramus
	The massive jaw indicates the presence of powerful jaw muscles	

Neanderthal Man - *Homo neanderthalensis*

During the year 1848-1861, the residues of the Neanderthal man from the late Pleistocene bed (before or during the first Ice Age) were found in Spain and North Africa to Ethiopia, Mesopotamia., Gibraltar, Neanderthal Valley near Dusseldorf (Germany), Southern Russia.

They possess characteristics features as mentioned in Table 13.5.

Table 13.5: Characteristics features of *Homo neanderthalensis*.

Sr. No.	Organ/body part	Characteristics feature
1.	Skull	Massive long and flat-topped.

2.	Forehead	Receding
3.	Brow Ridges	Heavy
4.	Nose	Broad
5.	Orbits	Large
6.	Jaws	Protruding
7.	Chin	Receding
8.	Teeth	Large
9.	Average brain volume	1450 c.c.
10.	The attachment sites of the occipital region of the skull and the cervical vertebrae indicate the existence of powerful neck muscles.	

The Neanderthal man lived around 100,000 years ago. The males were bigger than the females. Their residence was in caves and rock shelters made with stone tools and weapons. There were also indications of the use of fire. Human evolutionary tree is shown in Fig. 13.1.

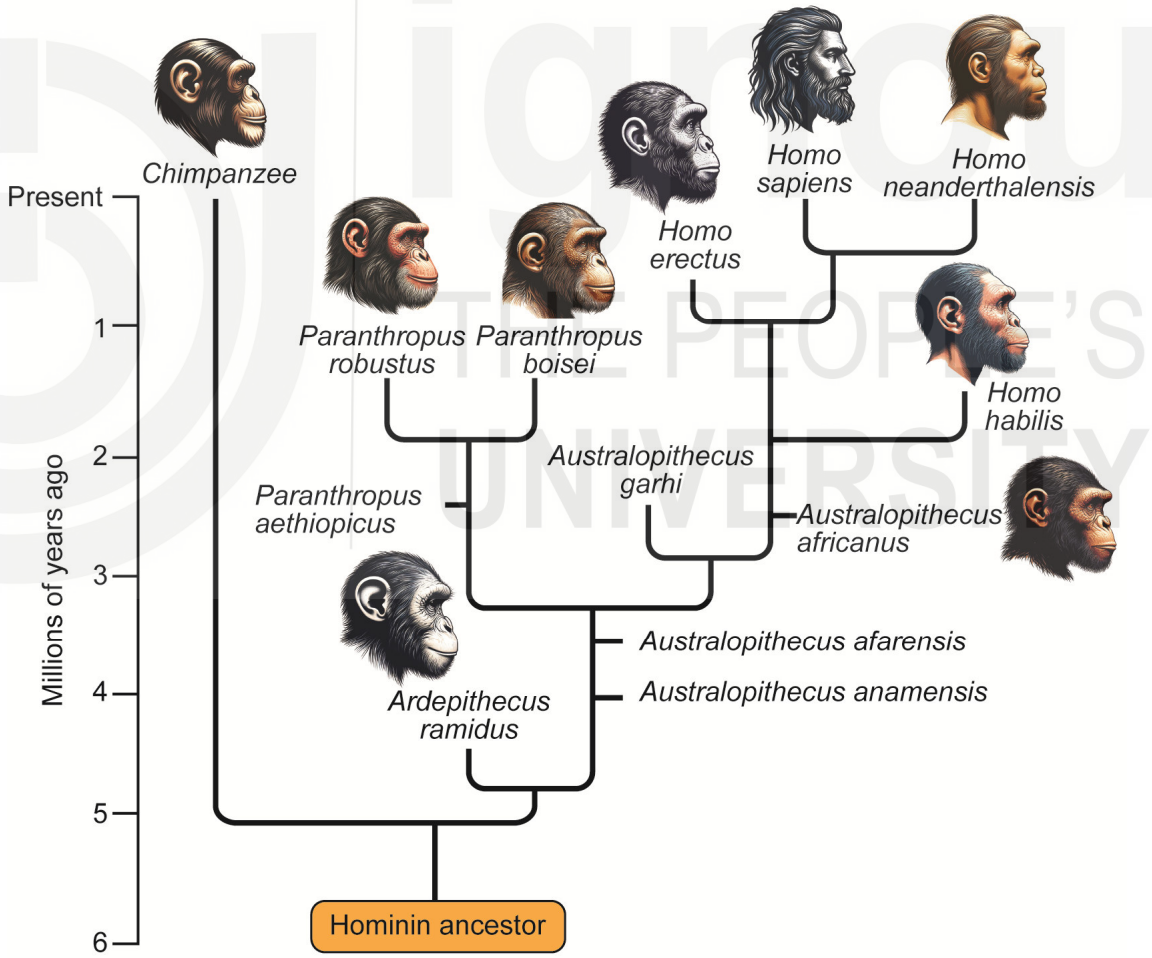

Fig. 13.1: Human evolutionary tree.

13.3 TREE OF LIFE

Evolution does not proceed in a simple straight line, but takes the form of a vast tree-like path consisting of various diverging, and sometimes also converging, branches going in many different directions. This insight, that

evolution is tree-like, constituted a huge shift in understanding that still has not totally overthrown the old linear thinking in much of popular culture. The tree of life explains the relationships between the different species on Earth. From microorganisms to trees to fungi and animals, life has evolved through time down countless pathways to provide us with the present-day collection of different species. The common belief in biology is that all living things evolved from a common ancestor more than 4 billion years ago.

The branches of the tree of life are formed from different groups of organisms. Two branches that are close to each other contain closely related organisms. The first and largest branches from the tree of life are formed by three domains. The branches of each domain split into many more branches. Life is currently separated into three different domains: Bacteria, Archaea and Eukarya. Bacteria and Archaea, consist entirely of microscopic single-celled organisms. The third domain, Eukarya, includes many microscopic organisms but also contains higher groups such as animals, plants, and fungi.

Bacteria and Archaea are called prokaryotes because their genetic materials are not enclosed in a membrane. The organisms in the domain Eukarya have cells with a nucleus. It is an extremely diverse and variable domain. It includes thousands of microscopic organisms plus all the large animal and plant species that are found on land and in water.

Charles Darwin (1809–1882) used the metaphor of a "tree of life" to conceptualize his theory of evolution. In *On the Origin of Species* (1859) he presented an abstract diagram of a theoretical tree of life for species of an unnamed large genus. Ernst Haeckel (1834–1919) constructed several trees of life. His first sketch, in the 1860s, shows "*Pithecanthropus alalus*" as the ancestor of *Homo sapiens*. His 1866 tree of life from *Generelle Morphologie der Organismen* shows three kingdoms: Plantae, Protista and Animalia. This has been described as "the earliest 'tree of life' model of biodiversity". His 1879 "Pedigree of Man" was published in his 1879 book *The Evolution of Man*. It traces all life forms to the Monera, and places Man (labelled "*Menschen*") at the top of the tree.

In 1990, Carl Woese, Otto Kandler and Mark Wheelis proposed a "tree of life" consisting of three lines of descent for which they introduced the term **domain** as the highest rank of classification. They also suggested the terms bacteria, archaea and eukaryota for the three domains. The model of a tree is still considered valid for eukaryotic life forms.

Due to horizontal gene transfer within the prokaryotes at the single and multicell level, the tree of life does not explain the full complexity of the situation in the prokaryotes.

13.4 GENOMICS AND HUMANNESS

The work on Human Genome Project started in 1990 under which the nucleotide sequence of the entire human nuclear genome is being determined. The mitochondrial genome was sequenced in the early 1980s. The human genome has been the focus of biological research for the last decade and will continue to be the center of attention for many years to come.

The human gene catalog, containing a description of the sequence of every gene in the genome, will be of a great value. The catalog contains the sequences of the coding parts, non coding and regulatory part of the every gene. The human gene catalog will provide the information about the genetic defects and thus underlying basis to these diseases, leading to strategies for treatment and management. While the catalog is being completed, attention will focus more and more on the transcriptome and proteome, which are the keys to understanding how the information contained in the genome is utilized by the cell. The Human Genome Project, and the similar projects currently being carried out with other species' genomes, therefore opens the way to a comprehensive description of the molecular activities of human cells and the ways in which these activities are controlled.

The genome projects will have additional benefit. The human genome contains extensive amounts of intergenic DNA. We think that most of the intergenic DNA has no function, but perhaps this is because we do not know enough about it. The first step in addressing this possibility is to obtain a complete description of the organization of the intergenic DNA in different genomes, so that common features, which might indicate a role for some or all of these sequences, can be identified.

Furthermore, investigations of minisatellites and microsatellites linked to one-of-a-kind occurrences, such as deletions, have discovered that variation present beyond Africa is a subset of variation seen in Africa. The investigation of fossil DNA sequences would allow direct inference of the genetic link between extinct hominids and modern humans. According to these sequences, the mitochondrial genomes of modern humans and Neanderthals share a common ancestor about 500000 years ago but they are not prevalent in the human populations over the world. These findings show that Neanderthals did not contribute any mitochondrial DNA to the present human gene pool.

13.4.1 Markers of Humanness

Humans also can be compared to worms, mice or apes at a structural, genomic level. But, humans cannot be put in animals group in relation to other primates. Chimps share 98% of the genome with humans. Humans have 10,000 more genes than a worm. The genomes of the yeast and human have similarities up to large extent. Humans do not have more genes than other mammals. The meaning of genes and genomes is more important than the number of genes. Thus, human and non-human difference is constructed in terms of genetic quality. Humans have several genes which are better than the genes of the animals. Human's genes are more 'flexible' and 'developed. Function of the genes is important than the number of the genes. The chimpanzee is said to share 98% of its genome, as well as its behavioural traits and social systems with the human. Humans possess additional qualities than the chimpanzee such as language, ability to read and free will. The human thus encompasses the chimp (and other mammals).

The markers of humanness could consist of the ability to speak and communicate, make moral decisions, be aware of existential dimensions and take responsibility for the future of the planet. But the development of the culture becomes more and more important to define the particularity of the human. An individual's genome discloses his or her ancestral lineage, which is a function of the migrations and interbreeding among population groups. There is no race-specific gene in humans. Humans are the one race. Humans address issues of concern such as human rights, education, job opportunities, climate change and global health.

13.5 RECENT TRENDS IN HUMAN EVOLUTION

There are many trends observed in the remains of the hominid such as differences in the facial angle, cranial capacity and posture. Human's evolutionary history is reconstructed as a bush, with various related species present at any given time.

13.5.1 Trends in Cranial Capacity

The first hominids were supposed to have a body similar to that of ape with head similar to that of a modern man. Over the course of human evolution, cranial capacity has gradually increased. As a result, *Sahelanthropus* and the early *Australopithecines* possessed cranial capacities similar to modern chimps (about 400 cc on average), with the later *Australopithecines* reaching 550 cc. Early *Homo erectus* skulls had brain sizes of roughly 510 cc. Later specimens have brain sizes of up to 1225 cc.

13.5.2 Bipedalism

Bipedalism is a major type of locomotion, involving movement on two feet. Reconstruction of the fossil constructed with the help of computer based programmes reveals a foramen magnum beneath the cranium and a modest nuchal crest, implying bipedalism of this species. *Australopithecus afarensis*' pelvis, leg, and foot bones also imply the bipedalism of this species though it was not very well adapted for running. Further, the position of foramen magnum (which is halfway between that of apes and humans) shows that *Australopithecus afarensis* was not able to keep its head fully erect. Due to the narrower pelvic exit in erectus, it is possible that he was an even more efficient biped than the modern human. *Sapien*'s broader pelvic outlet, which allows for the birth of large-brained babies, sets the hip joints more apart than is necessary for optimal locomotory efficiency.

In *Homo sapiens*, another trait associated to bipedalism is the Achilles tendon that attaches the calf muscles to the tarsal bones of the heel. The calf muscle extends all the way down to the tarsal bones in large apes, but not in humans. Humans have an Achilles tendon, which makes them good endurance runners, and is also useful for active hunting on the open savannah. It acts as a spring if the tendon is stretched leading to the collection and release of the energy after the foot pushes off for the next step. By this mechanism 50% of the metabolic energy for fast locomotion can be saved.

SAQ 1

Fill in the blanks:

a) The only surviving species of the genus *Homo* is ……..

b) ……. is considered to be the earliest known ancestor of man.

c) ……. invented the fire and were carnivorous.

d) *Homo habilis* is said to be of intermediate stage between the …… and …….

e) Life is currently separated into three different domains that is ……, …….. and …….

f) Bacteria and Archaea are called ……. because their genetic materials are not enclosed in a membrane.

g) The domain ……. have cells with a nucleus and includes thousands of microscopic organisms plus all the large animal and plant species that are found on land and in water.

h) The human genome contains extensive amounts of …… DNA.

i) Over the course of human evolution, cranial capacity has gradually ……...

j) The …… is a major type of locomotion, involving movement on two feet.

13.6 TRENDS IN GENERAL MORPHOLOGY

Dental arcades

The teeth of an ape are placed in a rectangular dental arcade with two parallel lines of left and right cheek teeth. The dental arcades of *Australopithecus* are more rectangular than parabolic. The dental arcade of *Homo* species is broader at the back than the front and is a full parabola. A tendency in tooth size with modern human's cheek teeth is observed as smaller than *Australopithecine's.* The tooth size has decreased drastically over the last 30,000 years in *H. sapiens*.

Crests and ridges

The skulls of both large apes and early hominids exhibit prominent crests and ridges. The sagittal and nuchal crests, as well as the brow ridges, are the most visible. The sagittal crests provide support for massive chewing muscles when present, and are consequently most prominent in species whose diet consists of hard, rough food that requires a lot of chewing. They're also bigger in men than in women, demonstrating sexual dimorphism. Large brow ridges are useful to channel the enormous forces exerted on the skull by a diet of coarse vegetable matter. The presence of a pronounced nuchal crest, as well as the

position of the foramen magnum, offers information about a species' posture. In species where the skull is not balanced vertically atop the spine, the crest serves as an anchor for neck muscles. As a result, modern humans have a completely upright posture, with the foramen magnum in the centre of the head and no nuchal ridge. Early *Australopithecines* like "Lucy" (*A. afarensis* was a hominid-becoming-hominin species, dated at 4.4 million years ago) possessed a foramen magnum that is halfway between humans and apes, as well as a tiny nuchal ridge. This indicates that they were not totally upright.

Facial angle

With the development of more recent hominids, there has been a general trend toward a flatter facial angle, culminating in *Homo sapien's* vertical (orthognathous) face. However, even among our lineage's elder members, there is tremendous variation: The moniker "flat-faced ape-man from Kenya" comes from *Kenyanthropus platyops*, which means "flat-faced ape-man from Kenya."

Sexual dimorphism

Variations in size, the shape of the pelvis and the crests on the skull, may be observed in all hominoids. Male gorillas are roughly twice as heavy as females. In chimps, the size disparity is significantly smaller, and even less so in modern humans, where males are 1.2 times heavier than females. In hominid fossils, this tendency toward decreased sexual dimorphism can be observed. *Australopithecine* skeletons had a significant degree of sexual dimorphism that is lowered in early hominids.

Changes in size of ribcage

A barrel-shaped ribcage is found in *Homo erectus* and waist separates the ribcage from the hips. The presence of a waist and a barrel-shaped ribcage in *H. erectus* indicate that large amount of meat were consumed. Meat has far more calories and is easier to digest than an all-vegetable diet. An omnivore, sometimes known as a carnivore, has a considerably shorter gut than a vegetarian. Consumption of higher meat with its higher fat content and calories may help to meet the higher energy demands of a larger brain. In *Homo erectus,* average cranial capacity increases dramatically.

13.7 UNIQUE PHENOTYPES OF MODERN HUMANS

Our understanding of the phenotypic and behavioural features that are modern human-specific is far more limited than the general list of features distinguishing us from other living apes. Many of which were shared by extinct hominins. The high and rounded forehead, presence of a chin, lack of brow ridges, gracile skeleton, and a distinct shape of the clavicle. It has been argued that many of the cranial features of modern humans are associated with their generally less robust skeleton and that the package of these gracile traits in the cranium may relate to a reduction in the length of a single element that is the sphenoid which is the central bone of the cranial base. The differences in human and Neanderthal clavicles, on the other hand, suggest

functionally different shoulder architectures that enable humans to raise their arms to the extent that allows them not only to climb, but also to throw, carry, and manipulate heavy objects.

Compared with this consensus on the package of traits defining the anatomical modernity of humans, the package of traits defining behavioural modernity has been more debated. Our grasp of the morphological and behavioural characteristics that distinguish contemporary humans from other living apes, many of which were shared by extinct hominins, is significantly more limited than the generic list of qualities that divide humans from other living apes. Omo I, the earliest known (195 KYA) anatomically modern human fossil, exhibits various traits with present human populations that distinguish it from previous forms of *Homo*, particularly *H. heidelbergensis* and Neanderthals, our closest cousins. In comparison to the consensus on the set of qualities that define human physical modernity, the set of traits that define behavioural modernity has been more contentious. Symbolic thinking, personal ornamentation, art, and long-distance trading of raw materials were once thought to be the hallmarks of our species, as evidenced by material evidence in the archaeological record. However, many of the components of this package have now been discovered in the remains of other hominins, such as Neanderthals. Instead than being diagnostic for "archaic" *versus* "contemporary" human behaviour, such characteristics could be thought of as reflecting the behavioural diversity of a variety of hominin species.

13.7.1 Genetic Uniqueness of Humans and Hominins

Although the human genome differs from that of other great apes by only a few percent, there are millions of structural and single nucleotide variations. Most of them are unlikely to have any phenotypic significance, but a few are likely to be linked to human-specific physical and behavioural traits, regardless of whether natural selection has influenced their evolution. Following the publishing of the chimp, orangutan, and gorilla reference sequences, genome-wide comparisons of humans and other primates became possible. The release of the Neanderthal and Denisovan genomes has opened up the potential of discovering the genetic modifications that distinguish humans from them. We can now distinguish between mutations that our forefathers shared with these extinct hominin species and those that are really unique to humans.

Humans and other great apes have sequence and structural differences. There are around 40 million variations between the human and chimp genomes, with 35 million nucleotide changes and 5 million indels. Humans share all but a few of their genes with apes, and the formation of new ape genes from noncoding sequence is regarded both highly unusual and difficult to find because it requires reliable orthologous sequence data from nonhuman species. More than 60 novel human genes have been identified since the publication of human and great ape genome sequences. These genes are primarily expressed in the cerebral cortex and testes, although this is true for many other genes. Furthermore, the novel genes are typically short, and their functional role in human phenotype uniqueness is unknown. We would expect to see rare back-mutations if we turned the original ape's phenotypic package into a human one; yet, none of the known mutant human phenotypes result in an ape-like phenotype. This line of reasoning, however, does not tell us if

there have been 100, 1000, or 10,000 functionally significant changes. Many human specific phenotypes, such as the opposable thumb and white covering of the eyeball, are likely to have completely different genetic causes and have evolved along separate evolutionary paths, but many other traits, such as brain development and culture-defining traits, may share some common genetic basis.

Given the significant sequence similarity between human and chimp genomes, it was assumed that regulatory mutations, or those that alter the transcription pattern of specific genes, would be the most important in explaining human phenotype diversity. If these mutations impact genes that encode transcription factors, which regulate other genes, a single mutation may affect a huge number of genes indirectly. To explain the success of anatomically modern humans, anthropologists have sometimes favoured hypotheses assuming the possibility of specific mutations with substantial effects directing the evolution of essential human features such as brain polarity or language. Although most human-specific phenotypes are complicated and polygenic, there may have been genetic changes that had a significant impact on the development of human and hominin-specific features. Body size in dog breeds is mostly governed by a polymer prism in a single growth factor gene, IGF1.67, which is a well-known example from another species. Phenotype differences can also be caused by structural alterations in the genome, such as gene duplication or deletion. The "more is better" viewpoint in vertebrate genomics has underlined the importance of gene duplications in evolution, owing to the fact that many duplicated genes are maintained after whole genome duplication events because they can obtain new functions. Another point of view is that humans are "degenerate apes" in some ways, and that several of our unique characteristics such as sluggish development, hair loss, and muscle weakness could be due to loss-of-function mutations. All of the mutation classes have the potential to contribute to significant ape-human differences. Tandem repeated DNA sequences evolve quickly; in fact, the blocks of heterochromatin near the centromeres and telomeres are the most rapidly changing sections of the genome. Because of their low gene density, they are poor candidates for functionally significant alterations. Interspersed repeat differences, such as SINE, LINE and retrovirus-derived long terminal repeats (LTRS), can, on occasion, have functional repercussions. The human-specific endogenous retrovirus family HERV-K cluster 9 and LTRI are examples of distinct LTRS. When inserted into genes, interspersed repeats have influenced human phenotype evolution by inactivating genes and modulating gene expression.

On the human lineage, segmental deletions and duplications affect more base pairs than SNPS, and are enriched with genes involved in reproduction, immunity, and chemosensory activity. The low quality of their genomic sequences in duplicated regions has complicated fine-scale comparisons with great ape genomes. Even when considering genomes that have been accurately sequenced, the rate of gene loss and gain in humans and great apes is more than double that of other animals. In addition, numerous gene families in great apes demonstrate a growth of copy number that is only seen in humans. *SRGAP2*, is one among these. Humans and chimps have a twice larger number of gene deletions than macaques, and a few genes that are active in apes and other mammals are inactive in humans. The mandible-expressed *masticatory myosin heavy chain 16* gene (*MYH16, OMIM 608580*)

has become inactivated in humans due to a frameshift mutation dated to 2.4 MYA. The Alu-mediated inactivation of the *cytidine monophospho-N acetylneuraminic acid hydroxylase* gene (*CMAH*, *OMIM 603209*), which governs the cell-surface distribution of sialic acid types is important in cell-cell communication, is another example of human-specific gene loss. As a result, human cells are deficient in N-glycolylneuraminic acid (Neu5Gc), with trace levels seen in the diet. The enzyme's inactivity is caused by a chromosomal loss that eliminates a 92-bp exon and causes a frameshift, resulting in a truncated protein. It's likely that this shift was caused solely by genetic drift, and that it has no bearing on human phenotype. However, because the gene is present in most other species and is involved in a range of biological processes, it is possible that its deletion was chosen for. As many viruses bind to these sugars on the cell surface, the loss of Neu5Gc could have given a means for past populations to resist a pathogen. Sialic acids, such as Neu5Gc, have been shown to preserve in fossils better than DNA, and because the lack of Neu5Gc is shared by contemporary humans and Neanderthals, the gene's inactivation is believed to predate their common ancestor -500 KYA. Another distinction between ape and human genomes may be the inactivation of gene. SIGLEC12, a protein found in apes, interacts to Neu5Gc and requires an usually conserved arginine within the polypeptide. SIGLEC12 is found in humans, but the human version lacks the arginine required for Neu5Gc binding. SIGLEC12's function in humans is unknown. There appears to be a group of functionally related genes co-evolving in a certain pattern on the human lineage since it has lost or significantly modified its activity.

More than ten changes in *siglec* (sialic acid-binding immunoglobulin superfamily lectins) genes have been identified as being uniquely human, including changes in enzyme binding efficiency, gene expression pattern, sequence change through gene conversion, and deletion or inactivation, implying that sialic acid biology has been a hotspot of human evolution. Some of the highly variable deleted *siglec* genes have functional relevance to immune and neurological tissue in humans. A genome-wide scan found 510 sites with human-specific deletions of regulatory sequences that are otherwise conserved among animals. One of these was shown to be close to the tumour suppressor gene *GADD45G*, whose deletion is linked to the proliferation of particular brain areas. A second case, a deletion of the regulatory region of the *androgen receptor* gene (*AR*, *OMIM 313700*), has been associated with the loss of facial vibrissae and penile spines in the human lineage. Interestingly, some of the genes, like the *immunity-related GTPase family M* gene (*IR*GM, OMIM 608212), have been lost in ancestral primates and been "resurrected" in the human lineage.

SAQ 2

Fill in the blanks:

i) The dental arcade of ……. species is broader at the back than the front and is a full parabola.

ii) In hominid fossils, this tendency toward …… sexual dimorphism can be observed.

iii) A barrel-shaped ribcage is found in …….. and waist separates the ribcage from the hips.

iv) More than ……. novel human genes have been identified since the publication of human and great ape genome sequences.

v) The low gene density in the …….. makes them poor candidate for functionally significant alterations.

vi) Sialic acids such as …….. have been shown to preserve in fossils better than DNA.

vii) The earliest known anatomically modern human fossil named as …… exhibits various traits with present human populations that distinguish it from the closest cousins ……. and ……..

13.8 SUMMARY

- Humans are *Homo sapiens*, an upright walking species that developed about 315000 years ago on the ground.
- Humans were preceded by other hominins such as *Ardipithecus, Australopithecus*, and other *Homo* species by millions of years, according to abundant fossil evidence.
- Some evolutionary models consider *Graecopithecus* to be ancestral to the human lineage, which are *Australopithecus, Paranthropus*, and *Homo,* while others consider *Graecopithecus* to be close to great ape ancestry, which includes Chimps, Bonobos and Gorilla.
- The tree of life increased as more varieties occurred, and some organisms became more mobile and developed a mouth that opened into a stomach, while others developed a body.
- *Australopithecus afarensis* of East Africa evolved into the earliest humans 3 million years ago.
- Forefathers of humans crossed the Rubicon of brain size 2.5 million years ago. *Homo habilis* was the first member of *Homo* genus.
- From a molecular-genetic standpoint, it's apparent that the DNA sequences found in today's people have been passed down from prior generations. The mitochondrial genome is where the most information on human DNA sequence diversity is currently available. The variance in these DNA sequences has a chronological depth of 500000 to 1.9 million years. Neanderthals did not contribute any mitochondrial DNA to the present human gene pool.
- Several trends in human evolution can be seen in hominid remains, including variations in posture, cranial capacity, and facial angle. Furthermore, the brain size of hominids, especially *Homo* species, is larger than would be expected for animals of their size.

- *H. sapiens*, tooth size has shrunk dramatically over the last 30,000 years. The skulls of both large apes and early hominids exhibit prominent crests and ridges. The presence of a pronounced nuchal crest, as well as the position of the foramen magnum, offers information about a species' posture.
- In *Homo erectus*, average cranial capacity increases dramatically and other morphological features had reduced sexual dimorphism, changes in size of ribcage.
- There are around 40 million variations between the human and chimp genomes, with 35 million nucleotide changes and 5 million indels. Given the significant sequence similarity between human and chimp genomes, it was assumed that regulatory mutations, or those that alter the transcription pattern of specific genes, would be the most important in explaining human phenotype diversity. There appears to be a group of functionally related genes co-evolving in a certain pattern on the human lineage since it has lost or significantly modified its activity.
- A genome-wide scan found 510 sites with human-specific deletions of regulatory sequences that are otherwise conserved among animals. One of these was shown to be close to the tumour suppressor gene *GADD45G*, whose deletion is linked to the proliferation of particular brain areas. A second case, a deletion of the regulatory region of the *androgen receptor* gene (*AR*, *OMIM 313700*), has been associated with the loss of facial vibrissae and penile spines in the human lineage.
- Interestingly, some of the genes, like the immunity-related GTPase *family M* gene (*IRGM*, *OMIM 608212*), have been lost in ancestral primates and been "resurrected" in the human lineage.

13.9 TERMINAL QUESTIONS

1. Describe the history of human evolution with a help of suitable diagram.
2. What is a “Tree of life”? Explain.
3. Correlate the genomics and humanness with reference to the evolution of humans.
4. Explain the change in the human cranial capacity occurred during the process of evolution?
5. Explain “Bipedalism” with respect to human evolution.
6. Explain the different morphological changes occurred in human during the process of the evolution.
7. Describe the phenotypes that are unique to modern humans.
8. What genes are deleted/ mutated during the process of human evolution and what is their significance?

13.10 ANSWERS

Self-Assessment Questions

1. a) *Homo sapiens*
   b) *Dryopithecus*
   c) *Homo erectus*
   d) *Australopithecus*; *Pithecanthropus erectus*
   e) Bacteria; Archaea; Eukarya
   f) prokaryotes
   g) Eukarya
   h) intergenic
   i) increased
   j) Bipedalism
2. a) *Homo*
   b) decreased
   c) *Homo erectus*
   d) 60
   e) heterochromatin
   f) Neu5Gc
   g) Omo I; *H. heidelbergensis*; Neanderthals

Terminal Question

1. Refer to Section 13.2.
2. Refer to Section 13.3.
3. Refer to Section 13.4.
4. Refer to Sub-section 13.5.1.
5. Refer to Sub-section 13.5.2.
6. Refer to Section 13.6.
7. Refer to Section 13.7.
8. Refer to Sub-section 13.7.1.

GLOSSARY

Adaptive radiation : Adaptive radiation is the rapid diversification of an ancestral population into several ecologically different species, associated with adaptive morphological or physiological divergence.

Allopatric speciation : It is also known as geographic speciation, is speciation that occurs when biological populations of the same species become isolated due to geographical changes such as mountain building or social changes such as emigration.

Allopolyploidy : It arises by multiplication of chromosome sets resulting from intergeneric or interspecific hybridisations.

Allozymes : These are the different molecular forms of an enzyme that correspond to different alleles of a common gene (locus).

Alternation of generations : It means that plants alternate between two different life stages, or generations, in their life cycle; a haploid stage called gametophyte and a diploid stage called sporophyte. The terms haploid and diploid refer to the number of chromosomes contained in the cells.

Alternative splicing : It is a cellular process in which exons from the same gene are joined in different combinations, leading to different, but related, mRNA transcripts. These mRNAs (all from a single gene) can be translated to produce different proteins with distinct structures and functions.

Anagenesis : The evolutionary process whereby one species evolves into another without any splitting of the phylogenetic tree.

Apicoplast : It is the relict of a plastid organelle found in several disease-causing apicomplexan parasites such as *Plasmodium* spp. and *Toxoplasma gondii*. In these organisms, the organelle has lost its photosynthetic capability but harbours several fitness-conferring or essential metabolic pathways.

Archipelago : It is an area that contains a chain or group of islands scattered in lakes, rivers, or the ocean.

Artificial classification : It is a method of grouping organisms based on non-evolutionary characteristics, such as physical traits.

Artificial speciation : It is the process of creating new species through human intervention, such as selective breeding.

Autopolyploidy : It is a condition where an organism has more than two sets of chromosomes from the same species.

Biodiversity : The Convention on Biological Diversity defines biodiversity as the "variability among living organisms from all sources".

Biosphere : It is the region of the earth that encompasses all living organisms (plants, animals and bacteria).

Biosystematics : It is the taxonomy based on the study of the genetic evolution of plant and animal populations.

Bipedalism : The ability to maintain a standing position, and to move about, using only two legs.

Birth-and-death model of evolution : In this model of evolution, new genes are created by repeated gene duplication and some duplicate genes are maintained in the genome for a long time, but others are deleted or become nonfunctional by deleterious mutations.

Bottleneck effect : It is a dramatic reduction in the size of a population, which can lead to a loss of genetic variation. This can happen due to environmental events like earthquakes, floods, and fires, or human activities like genocide or culling.

Bradytelic evolution : An exceedingly slow rate of evolution, manifest by slowly evolving lineages which survive much longer than would normally be expected.

Calmodulin : It is a protein with four calcium-binding sites.

Centromeres : These are the chromosomal regions that become physically linked to the spindle during cell division; ensure equal division of genetic material between daughter cells.

Chondrogenesis : It is the initial development of skeletal bones, starts during embryonic development in the early stages of somitogenesis and is considered to be the earliest phase of skeletal development.

Clade : A group of two or more taxa or DNA sequences that includes both their common ancestor and all their descendent

Cladogenesis : It is the division of a single lineage into two lineages.

Cladogram : It is an evolutionary tree that diagrams the ancestral relationships among organisms.

Clasper : It is an intromittent organ inserted into the cloaca and oviduct of the female during copulation.

Coelom : It is a body cavity filled with fluid that is found in animals and is located between the intestinal canal and the body wall.

Concerted evolution : It describes an unusual evolutionary pattern exhibited by certain repetitive DNA sequences. It is traditionally defined as a situation where repeats within a genome (paralogs) show more sequence similarity to each other than they do to their counterparts in other species (orthologs).

Cytokinesis : It is the physical process of cell division, which divides the cytoplasm of a parental cell into two daughter cells.

Dioecious : It refers to having the male and female reproductive organs in separate individuals (plant or invertebrate animal).

Ecological niche : It is defined as the specific set of environmental conditions required by an organism or the functions it performs in nature.

Ecomorphs : A small difference in form or colour that distinguishes populations of a species that have become reproductively isolated recently.

Embryogenesis : It is the process of development of an embryo from zygote.

Endocytosis : It is an energy-dependent process by which a cell internalizes extracellular substances or materials on the cell membrane by engulfing them.

Endothermy : In extant animals, it is defined by high metabolic rates that result in sufficient heat production to permit alteration of internal body temperature when the organism is at rest.

Enhancers : These are classically defined as cis-acting DNA sequences that can increase the transcription of genes.

Epigenetic modification : It refers to a reversible and heritable change in gene expression in the absence of a change in the DNA sequence in the nucleus; this modification includes chromatin remodeling, DNA methylation, and histone modification.

Epithet : It is the second part of the binomial name of a particular species.

Eukaryogenesis : It is the process that led to the creation of eukaryotic cells, which are more complex than prokaryotic cells and are the basis for all complex cells and almost all multicellular organisms.

Evolutionary innovation : It refers to a pattern of change defined by four principles, and the changes are the result of interactions among individuals, populations, and environments over time. An evolutionary pattern of technical change is here defined to include generation of novelty leading to diversity, retention and transmission of characteristics over time within the population which innovates, social selection, and an assumption of non-optimization.

Exocytosis : It is the process by which cells excrete waste and other large molecules from the cytoplasm to the cell exterior.

Exon shuffling : The creation of new genes by bringing together, as exons of a single gene, several coding sequences that had previously specified different proteins or different domains of the same protein, through intron-mediated recombination.

Founder effect : It is the reduction in genetic variation that results when a small subset of a large population is used to establish a new colony.

Gastrulation : It is an early developmental process in which an embryo transforms from a one-dimensional layer of epithelial cells, a blastula, and reorganizes into a multilayered and multidimensional structure called the gastrula.

Gradualism : It is a theory that assumes large morphological changes in organisms occur *via* a number of smaller step over a number of years.

Haplotype : It refers to a set of DNA variants along a single chromosome that tend to be inherited together. They tend to be inherited together because they are close to each other on the chromosome, and recombinations between these variants are rare.

Holozoic : It describes the organisms that feed by ingesting complex organic matter, which is subsequently digested and absorbed.

Homeostasis : A state of balance among all the body systems needed for the body to survive and function correctly.

Hominids : A primate of a family (*Hominidae*) which includes humans and their fossil ancestors and also (in recent schemes) at least some of the great apes.

Hominins : They belong all organisms, extinct and extant, within the human lineage after the split from the ancestors of the great apes.

Homoploid hybrid speciation : It is the formation of a new-hybrid species, independent from its parents, *via* hybridization with no whole-genome duplication and thus no increase in ploidy.

Hydrogenosomes : These are the anaerobic mitochondria that have a double membrane and synthesize ATP *via* hydrogen-producing fermentations.

Indels : It refers to the insertion or deletion of one or more nucleotides in one genome relative to another, which can be used as a sequence feature to characterize the evolution of different organisms.

Innovation-amplification-divergence (IAD) : In this model for evolution of new functions following gene duplication, the new function appears before duplication and functionally distinct new genes are evolve under continuous selection.

Karyogamy : It is the process of fusing the nuclei of two haploid cells during fertilization.

Karyotype : It is an individual's complete set of chromosomes. The term also refers to a laboratory-produced image of a person's chromosomes isolated from an individual cell and arranged in numerical order. A karyotype may be used to look for abnormalities in chromosome number or structure.

Living fossil : It is as an extant species that belongs to an old lineage that has undergone slow rates of evolution.

Metamerism : It is the phenomenon of having a linear series of body segments that are similar in structure but not necessarily identical. These segments are called metameres or somites in animals.

Microsatellites : These are the repeating sequences of 2–6 base pairs of DNA.

Minisatellite : It is a tract of repetitive DNA in which certain DNA motifs (ranging in length from 10 to 60 base pairs) are typically repeated 5–50 times.

Mitosomes : The mitosome is an organelle found in "amitochondrial" unicellular organisms which do not have the capability of gaining energy from oxidative phosphorylation. Unlike mitochondria, Mitosomes do not contain any DNA.

Myotome : A myotome is defined as a group of muscles which is innervated by single spinal nerve root.

Natural selection : It is a process in which an organism adapts to its environment through selectively reproducing changes in its genotype.

Neoangiogenesis : It is the development of new blood vessels from a pre-existing vasculature involves the migration behavior, proliferation and differentiation of endothelial cells. This process is an important aspect to both the growth and metastasis of solid tumors.

Neocortex : It is a complex brain structure that commands higher functions, such as sensory perception, emotion, and cognition.

Neofunctionalization : It is the mechanism explaining the origin of novel functions through gene duplication.

Neurogenesis : It is a developmental phenomenon defined as the generation and development of new functional neurons in the adult central nervous system (CNS).

Non-synonymous mutations : A non-synonymous mutation is nucleotide substitution that alters the amino acid sequence of a protein and is frequently subjected to natural selection.

Operculum : It refers to a body part that acts as a lid in order to protect another sensitive part of the body of a biological organism.

Oviparous : Animals that reproduce by laying an egg, which in turn will hatch into a juvenile organism.

Paleontology : Paleontology is the study of the history of life on Earth as based on fossils.

Paleontologists : They are the scientist who specializes in the study of life forms that existed in previous geologic periods, as represented by their fossils.

Palynology : It is the scientific discipline concerned with the study of plant pollen, spores, and certain microscopic planktonic organisms, in both living and fossil form.

Panmictic population : A panmictic population is one where all individuals are potential partners.

Paralogous genes : These are the genes present in a particular organism that are related to each other through a gene duplication event.

Parapatric speciation : It is a type of speciation that occurs when a species' population is separated into two subpopulations that evolve reproductive isolation from each other, while still exchanging genes.

Parthenogenesis : It is a reproductive strategy that involves development of a female (rarely a male) gamete (sex cell) without fertilization.

Pelagic species : These are the organisms that live in the water column of an ocean, lake, or coastal area, but not near the bottom or shore. The term "pelagic" can also refer to animals that live near the surface of the sea or forage at different depths.

Peripatric speciation : New species formed from a small population isolated at the edge of a larger population.

Phagocytosis : It is a process by which certain living cells called phagocytes ingest or engulf other cells or particles.

Phenogram : It is a diagram depicting taxonomic relationships among organisms based on overall similarity of many characteristics without regard to evolutionary history.

Pheromone : It refers to any endogenous chemical secreted in minute amounts by an organism to elicit a particular reaction from another organism of the same species.

Plasmodesmata : These are the specialized, intercellular channels that establish cytoplasmic and endomembrane continuities between neighboring cells in plants.

Plasmogamy : It is the fusion of the cytoplasm of two or more cells, usually gametes or protoplasts, without the fusion of nuclei.

Poikilotherms : Organisms with variable body temperature.

Porphyrin : A porphyrin is a large ring molecule consisting of 4 pyrroles, which are smaller rings made from 4 carbons and 1 nitrogen.

Proboscis : It refers to an elongated or projecting appendage in the head region of certain animals.

Proto-organelle : It is a lipid-encapsulated compartment that is constructed within a synthetic cell.

Pseudogamy : A form of sperm-dependent parthenogenesis in which eggs require activation by entry of sperm, but only maternal chromosomes are expressed and passed on.

Pseudogenes : These are the gene copies that have coding-sequence deficiencies like frameshifts and premature stop codons but resemble functional genes.

Punctuated equilibria : It is a hypothesis holding that the evolution of species proceeds in a characteristic pattern of relative stability for long periods of time interspersed with much shorter periods during which many species become extinct and new species emerge.

Reproductive isolation : It is the inability of different species to breed and produce viable offspring due to genetic, physiological, behavioral, or geographical barriers.

Retrotransposons : These are the mobile genetic elements present in all vertebrates, and which replicate by reverse transcription and insertion of cDNA into their host genome.

Sexual dimorphism : It refers to the differences in appearance between males and females of the same species, such as in colour, shape, size, and structure.

Spliceosomal introns : These are found in most eukaryotic genes which are non-coding sequences excised from pre-mRNAs by a special complex called spliceosome during mRNA splicing.

Splicing : The removal of introns and the joining of exons from eukaryotic primary RNA transcripts to create mature RNA molecules of the cytoplasm.

Stasigenesis : It refers to the persistence of an evolutionary lineage over time without any significant change or splitting. Living fossils are an example of stasigenesis.

Statocysts : An organ of equilibrium found in usually aquatic invertebrates that is typically a fluid-filled vesicle lined with sensory hairs which detect the position of suspended statoliths.

Subfunctionalization : It is the division of function such that the two members of a pair of the gene confer only part of the functions of the progenitor singleton.

Sympatric speciation : It refers to a type of speciation that occurs when two groups of the same species live in the same geographical area but no longer interbreed.

Synapsis : It refers to the pairing of homologous chromosomes, one from each parent, during early meiosis.

Syngamy : It is the fusion of two cells, or of their nuclei, in reproduction.

Systematics : It is the study and classification of organisms with the goal of reconstructing their evolutionary histories and relationships.

Tachytelic evolution : A rate of evolution within a group that is much faster than the average (horotelic) rate.

Taxonomy : It is the science of naming, describing and classifying organisms and includes all plants, animals and microorganisms of the world.

Telomeres : It can be defined as the heterochromatic regions found at the ends of chromosomes which essentially constitute repetitive DNA sequences (TTAGGG repeats).

Transcription factors : These are the proteins that regulate transcription by binding to specific DNA sequences and either promoting or inhibiting the process.

Vicariance : It is the geographical separation of a population, typically by a physical barrier such as a mountain range or river, resulting in a pair of closely related species.

Viviparous : Animals in which females gestate developing eggs inside their reproductive tracts, and give birth to their young.

Zooplankton : It pertains to the animal or animal-like organisms comprising the plankton.

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