Origin of life on earth

THEORIES FOR THE ORIGIN OF LIFE

Pasteur showed in an experiment in the nineteenth century that spontaneous generation of life from inorganic matter does not now take place – cells can only be formed from other cells. This is not surprising, as even the simplest prokaryotic cells are very complicated. Nevertheless, about 3.5 – 4.0 billion years ago, the first living cells did appear on Earth.

- Followers of many religions believe that God created life. This is called **special creation**.
- Some scientists believe that organic material can travel throughout the universe and so could have arrived on Earth from outer space. This is called **panspermia**.

The scientific method cannot be used to test these theories for the origin of life. Modern experiments can never establish with certainty what did or did not occur billions of years ago. They can only show what is possible, in conditions similar to those when life appeared. Results of experiments simulating conditions in interstellar dust clouds were published in 2001. Simple compounds such as ammonia, carbon dioxide and methanol were mixed with ice crystals in a vacuum at extremely low temperatures and in ultra violet light. Hundreds of complex organic compounds were formed, which self assembled into spherical vesicles. The results greatly strengthen the case for panspermia.

Another theory is that life did evolve here on Earth from inorganic matter, despite the fact that it is not possible today. In 1953, Stanley Miller and Harold Urey investigated this theory by trying to recreate the conditions of pre-biotic Earth. Inside their apparatus (top right) they mixed the gases ammonia, methane and hydrogen to form a reducing atmosphere. Electrical discharges and the boiling and condensing of water simulated lightning and rainfall. After one week, the clear water in the apparatus had turned to a murky brown. Analysis revealed many organic compounds, including 15 amino acids. Miller and Urey concluded that organic compounds could have formed on pre-biotic Earth.

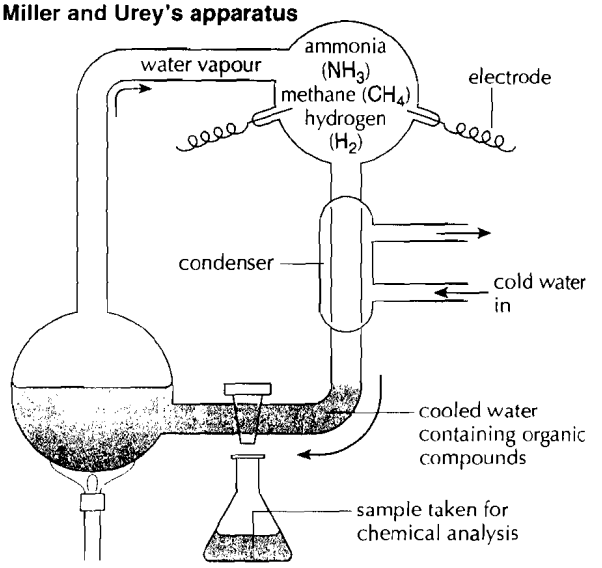
THE ORIGIN OF PROKARYOTIC CELLS

If prokaryotic cells did evolve from inorganic matter billions of years ago, membranes would have been needed. Phospholipids naturally group together to form bilayers. These membranes form into cell-like spheres, enclosing a droplet of fluid.

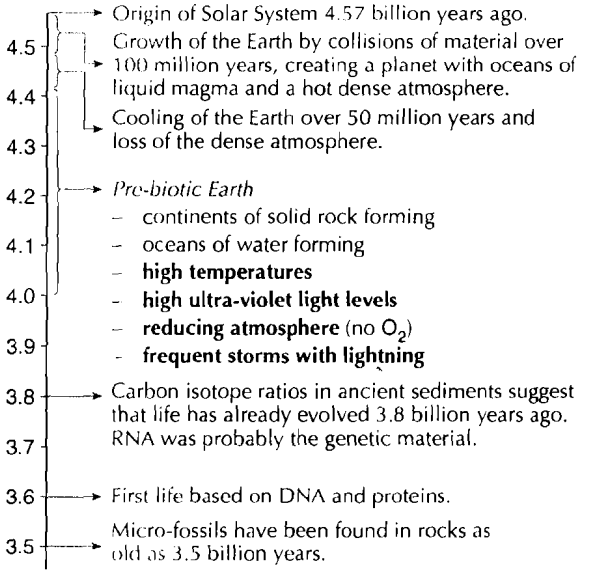
For these small structures to develop into living cells, a working genetic mechanism would have been needed. In modern prokaryotes the parts of the genetic mechanism cannot function without each other. For example, genes cannot be replicated without enzymes and enzymes cannot be made without genes. It seems inconceivable that the whole mechanism could have evolved at once, but gradual evolution would have required simpler intermediate stages. One possibility is the use of RNA instead of both DNA and enzymes. RNA can be replicated and can also act as a catalyst. Some reactions in the ribosome are still catalysed by RNA. Another possible intermediate stage is the use of clay minerals to catalyse reactions. Clay minerals are very variable. Some can divide, grow and catalyse specific reactions, including the formation of polypeptides from amino acids.

Before the origin of life, conditions on Earth were very different from those today. The figure (right) shows conditions on pre-biotic Earth.

Miller and Urey's apparatus



Conditions of pre-biotic Earth



THE ORIGIN OF EUKARYOTIC CELLS

Eukaryotic cells contain mitochondria and chloroplasts, which are not found in prokaryotic cells. If eukaryotic cells evolved from prokaryotic cells, the origin of these organelles must be explained.

According to the **endosymbiotic theory**, both mitochondria and chloroplasts have evolved from independent prokaryotic cells, which were taken into a larger heterotrophic cell by endocytosis. Instead of being digested, the cells were kept alive and continued to carry out aerobic respiration and photosynthesis. The characteristics of mitochondria and chloroplasts support the endosymbiotic theory.

- They grow and divide like cells.
- They have a naked loop of DNA, like prokaryotes.
- They synthesize some of their own proteins using 70S ribosomes, like prokaryotes.
- They have double membranes, as expected when cells are taken into a vesicle by endocytosis.
- Cristae are similar to mesosomes of prokaryotes
- Thylakoids are similar to structures containing chlorophyll in photosynthetic prokaryotes.

If species do evolve, a mechanism must exist to cause this evolution. Two theories have been proposed.

LAMARCK'S THEORY OF EVOLUTION

Lamarck, a French naturalist, proposed his theory in 1809. He observed that the characteristics of living organisms can change during their lifetime. For example, if muscles are used, they grow stronger. These are called acquired characteristics. Lamarck proposed that when organisms reproduce, they pass on these acquired characteristics to their offspring – this is called **inheritance of acquired characteristics**.

An example that is often discussed is the neck of the giraffe. The ancestors of the modern giraffe had short necks. According to Lamarck's theory they had to stretch up into trees to reach food, so their necks lengthened slightly. The next generation inherited the lengthened necks and stretched more. Many generations of this process would lead to the long neck of the modern giraffe.

Despite many attempts, no significant cases of inheritance of acquired characteristics have been found. Also, Lamarck's theory does not fit in with our knowledge of inheritance. For the characteristic to be inherited, acquired characteristics would have to cause a mutation in the gene controlling the characteristic. No mechanism for this exists.

DARWIN AND WALLACE'S THEORY OF EVOLUTION

Darwin and Wallace jointly proposed their theory in 1858. Darwin is usually given credit for it as he had been working on it for much longer and he published a detailed account of it in 1859. The Darwin–Wallace theory is evolution by natural selection. An explanation of this theory is given on page 38.

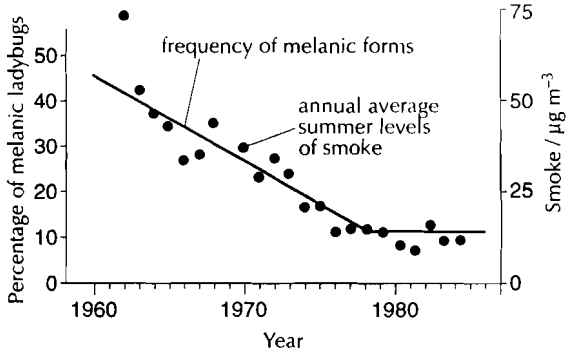
According to this theory, the ancestors of the giraffe varied in neck length. At times of food shortage, when all the lower leaves on trees had been eaten, only the giraffes with longer necks could reach the higher leaves. They survived and passed on their characteristics to their offspring, including longer necks. There is much evidence for evolution by natural selection, including modern examples of observed evolution.

MODERN EXAMPLES OF OBSERVED EVOLUTION

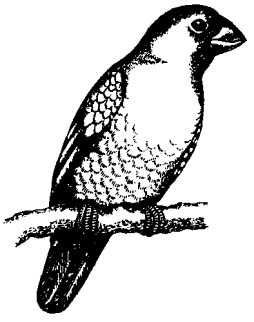
It is not realistic to expect large-scale evolution over a few years, but there are some well-documented examples of small-scale evolution.

1. Evolution of melanism in ladybugs

Adalia bipunctata, the two-spot ladybug (or ladybird) is a small beetle, which usually has red wing cases with two black spots. The red colour warns predators that it tastes unpleasant. Melanic forms also exist, with black wing cases. The melanic form absorbs heat more efficiently than the red form. It therefore has a selective advantage when sunlight levels are low and it is difficult for ladybugs to warm up. The melanic form of *Adalia bipunctata* became common in industrial areas of Britain, but declined again after 1960. The decline correlates with decreases in smoke in the air (below). In air darkened by smoke, the melanic forms will be able to warm up more quickly, but if the smoke is no longer present this advantage is lost and warning colouration is more important.



2. Evolution of the beaks of Galapagos Islands finches



Rosemary and Peter Grant have studied the two species of finch that inhabit Daphne Major, in the Galapagos Islands, for many years. One of the finches, *Geospiza fortis* (above), has a short, wide beak and feeds on a variety of seeds, including large hard ones. During 1982 – 3 there was a severe El Niño event, which brought very heavy rain to Daphne Major. With more food available, the population of *G. fortis rose* considerably, reaching a peak in 1983. It dropped back in the drier years following, and in 1987 was only 37% of its peak. The period of heavy rain changed the vegetation and until 1991 there were fewer plants producing large, hard seeds and more producing small soft ones. The diet of *G. fortis* therefore changed. The 37% of the population remaining in 1987 were not a random sample of the population. They had longer, narrower beaks than the average in 1983. There had been a significant change in the beaks of the population. The conclusion that this change was caused by natural selection due to the change in diet is supported by evidence from the other finch on the island, *Geospiza scandens*. Its population rose and fell in the same way as *G. fortis*, but neither its diet nor the size of its beak changed.

Evidence for evolution

Although it is not possible to prove using the scientific method that the organisms on Earth today are the result of evolution, there is much evidence that makes it very likely.

EVIDENCE FROM BIOCHEMISTRY

There are remarkable similarities between living organisms in their biochemistry.

- All use DNA (or RNA) as their genetic material.
- All use the same universal genetic code, with only a few insignificant variations.
- All use the same 20 amino acids in their proteins.
- All use left, and not right-handed amino acids.

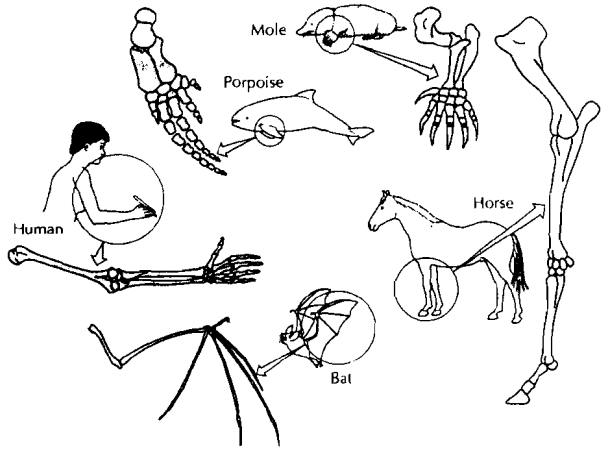
These similarities suggest that all organisms have evolved from a common ancestor that had these characteristics.

EVIDENCE FROM HOMOLOGOUS ANATOMICAL STRUCTURES

There are also remarkable similarities between some groups of organisms in their structure.

1. At an early stage, vertebrate embryos are very similar, despite huge differences in the structure of the adults (below).

2. The limbs of vertebrates show striking similarities in their bones, despite being used in many different ways (below). The structure is called the pentadactyl limb.

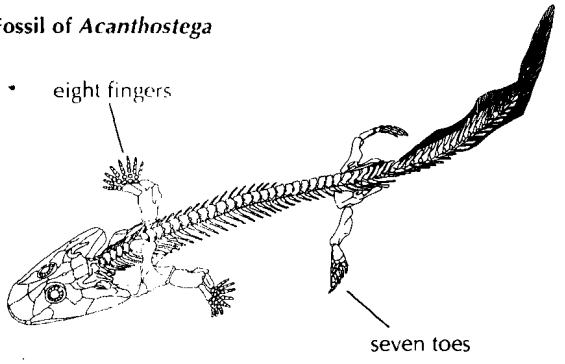


The most likely explanation for these structural similarities is that the organisms have evolved from a common ancestor. Structures that have developed from the same part of a common ancestor are called homologous structures.

EVIDENCE FROM PALAEONTOLOGY

The existence of fossils is very difficult to explain without evolution. An example of this is *Acanthostega*. The figure (below) is a drawing of a 365-million-year-old fossil of *Acanthostega*. It has similarities to other vertebrates, with a backbone and four limbs, but it has eight fingers and seven toes, so is not identical to any existing organism. This suggests that vertebrates and other organisms change over time. *Acanthostega* is an example of a 'missing link'. Although it has four legs, like most amphibians, reptiles and mammals, it also had a fish-like tail and gills and lived in water. This shows that land vertebrates could have evolved from fish via an aquatic animal with legs.

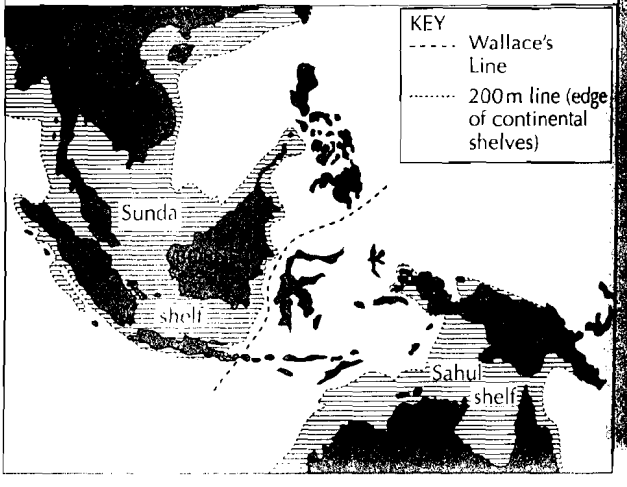
Fossil of *Acanthostega*



EVIDENCE FROM GEOGRAPHICAL DISTRIBUTIONS

Wallace's Line is an example of geographical distribution of organisms that is difficult to explain without evolution. The figure (below) shows its position. There are huge differences in the types of land animal that are found on either side of Wallace's Line. For example, placental mammals are found on the Asian side and mainly marsupial and monotreme mammals are found on the Australasian side. The landmasses on the two sides of the boundary separated about 100 million years ago and came together again by continental drift about 15 million years ago. The mammals on the separated landmasses followed different evolutionary paths, so different types evolved. In similar habitats where natural selection acts in the same way on different organisms, the results are sometimes strikingly similar, for example the marsupial mole of Australia and the golden moles of Africa.

Map showing Wallace's Line



The study of the evolutionary history of groups of organisms is called **phylogeny**. Two useful approaches in phylogeny are study of the fossil record and comparison of molecular structure.

DATING FOSSILS

To place fossils into a sequence, it is necessary to know their dates. Fossils, or the rocks containing fossils can be dated using radioisotopes – radioactive isotopes of chemical elements. When an atom of a radioisotope decays, it changes into another isotope and gives off radiation. The rate of decay varies between different radioisotopes and is expressed as the **half-life**. *The half-life is the time taken for the radioactivity to fall to half of its original level.* The figure (right) shows a decay curve for radioisotopes.

The two radioisotopes that are most commonly used are ^{14}C and ^{40}K. In radiocarbon dating the percentage of surviving ^{14}C atoms in the sample is measured. In potassium–argon dating, the proportions of parent ^{40}K atoms and daughter ^{40}Ar atoms are measured. In both methods the age in half-lives can then be deduced from the decay curve. The half-life of ^{14}C is 5730 years, so it is useful for dating samples that are between 1000 and 100 000 years old. The half-life of ^{40}K is 1250 million years so it is useful for dating samples older than 100 000 years.

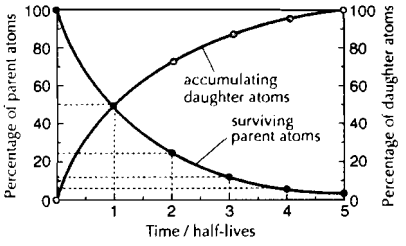
METHODS OF PRESERVATION

The remains of past living organisms can be trapped and preserved in various ways.

- In resins, which turn to amber.
- Frozen, in ice or snow.
- In acid peat, which prevents decay.
- In sediments that turn to rock.

The last method is the most important. Sediments accumulate in layers in parts of the sea and sometimes on land. The weight of sediments compresses those beneath until they become rock. If hard parts of animals such as shells or bones form part of the sediment, they will be preserved in the rock. Sometimes, the shape of an organism is preserved as a cast. Minerals sometimes seep into the soft parts of an organism as it decays and harden to form a petrified replica of the organism.

Decay curves for radioisotopes



COMPARING MOLECULAR STRUCTURE

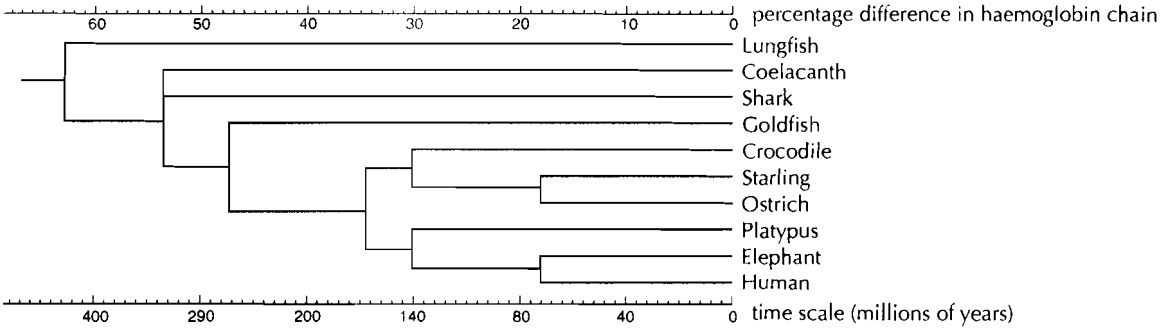
The phylogeny of many groups of organisms has been studied by comparing the structure of a protein or other biochemical that they contain. For example, the amino acid sequence of the polypeptide of hemoglobin has been compared in many vertebrates. The figure (below) shows the numbers of differences in the amino acid sequence in ten vertebrates. Differences in amino acid sequence accumulate gradually over long periods of time. There is evidence that differences accumulate at a roughly constant rate. They can therefore be used as an evolutionary clock. The number of differences in amino acid sequence can be used to deduce how long ago species split from a common ancestor. Using this information and the details of what the amino acid differences are, the probable phylogeny of groups of organisms can be deduced. The figure (bottom) shows the probable phylogeny of the ten organisms.

The phylogeny of many groups has been studied in this way. Usually, the results fit in with earlier studies of fossils, or anatomical studies, but sometimes there are surprises. For example, figure (bottom) shows larger than expected differences between the various types of fish.

Numbers of differences in the amino acid sequence of hemoglobin in ten vertebrates

	Elephant	Platypus	Ostrich	Starling	Crocodile	Lungfish	Coelacanth	Goldfish	Shark
Human →	26	40	43	41	47	83	70	68	71
Elephant →		45	45	48	50	84	72	63	74
Platypus →			54	52	51	89	74	70	76
Ostrich →				26	36	91	75	68	73
Starling →					47	91	77	67	70
Crocodile →						85	78	70	77
Lungfish →							90	94	86
Coelacanth →								83	78
Goldfish →									88

Phylogenetic tree diagram for ten vertebrates



Human origins

HUMAN CLASSIFICATION

Humans are classified as primates (see below), because they show primate characteristics:

- grasping limbs, with long fingers and a separated opposable thumb
- mobile arms, with shoulder joints allowing movement in three planes and the bones of the shoulder girdle allowing weight to be transferred via the arms
- stereoscopic vision, with forward facing eyes on a flattened face, giving overlapping fields of view
- skull modified for upright posture

These characteristics are sometimes described as adaptations for tree life, though many other tree-living mammals do not show them.

Human classification

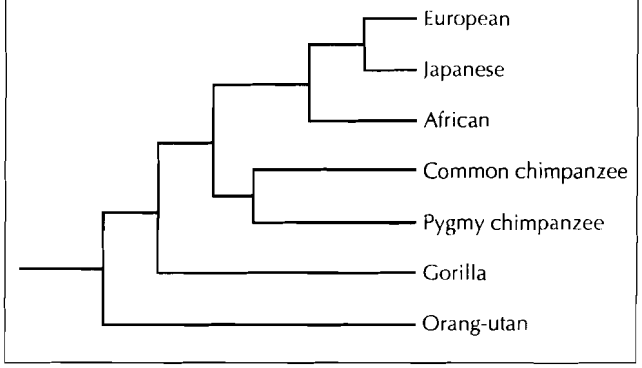
Kingdom	Animalia
Phylum	Chordata
Class	Mammalia
Order	Primate
Family	Hominidae
Genus	Homo
Species	sapiens
Subspecies	sapiens

SEARCHING FOR THE EARLIEST HUMAN ANCESTORS

The question of where the earliest hominid ancestors lived has still not been answered with certainty. The closest existing relatives of humans are chimpanzees and gorillas from Africa and orang-utans of South East Asia. Research into the differences between these primates in the amino acid sequences of their proteins, including hemoglobin, myoglobin and fibrinogen shows that humans are more closely related to chimpanzees and gorillas than orang-utans. The oldest human ancestors therefore probably lived in Africa.

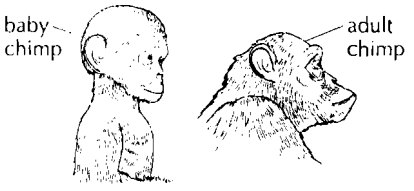
Mitochondrial DNA from humans and related primates has been sequenced. The differences in base sequence have been used to construct a hypothetical phylogeny (shown below). The data confirms that humans are closer to African apes than Asian ones and therefore supports the theory that human ancestors split from the ancestors of chimps and gorillas in Africa. The data also allows approximate dating of the splits between African and other humans – 140 000 ago and the split between Europeans and Japanese – 70 000 years ago. The conclusion for this and other studies is that the ancestors of modern humans migrated out of Africa less than half a million years ago. Other fossil hominids found out of Africa must have been the result earlier migrations and modern hominids are not descended from these hominids.

Phylogenetic tree for humans and closely related apes



NEOTENY

Neoteny is keeping juvenile characteristics as an adult. Adult humans show similarities in appearance to baby apes (below), with flat faces, large brain to body size ratio, upright heads and little body hair. This suggests that human evolution from an ape ancestor might have involved a slowing down of development, with a long childhood, delayed puberty and retention of juvenile characteristics in adulthood.



BIPEDALISM

Bipedalism is walking on two legs. Even the oldest fossils, of *Australopithecus afarensis*, show at least partial bipedalism, so it was a very early development in human evolution.

The change to bipedalism involved many adaptations.

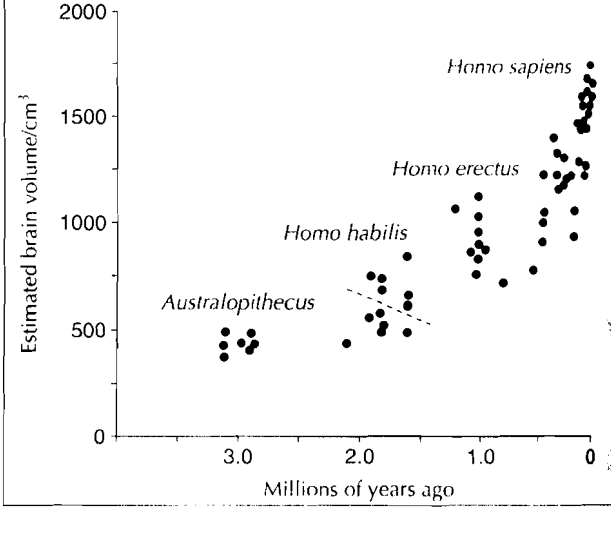
- The foramen magnum, a hole in the skull through which the spinal cord and brain connect, moved forwards. This allows the head to balance on the backbone.
- The arms became shorter and less powerful.
- The legs became longer and stronger.
- The knee changed to allow the leg to straighten fully.
- The foot became more rigid, with a longer heel, shorter toes and a non-opposable big toe.

There are many consequences of bipedalism. Collecting food from bushes is easier and also walking long distances while carrying food, water, infants, tools or weapons. It makes tree climbing more difficult.

INCREASED BRAIN SIZE

The brains of early hominids (*Australopithecus*) were only slightly larger in relation to body size than the brains of apes. The brains of later hominids (*Homo*) were larger (below). This was due to continued rapid brain growth after birth. In apes and earlier hominids brain growth slows after birth. There are many consequences of increased brain size. Capacities for learning, complex thought and memory are increased. Language and more complex tool manufacture and use are possible. However, the larger brains take longer to develop and more energy to use.

Brain sizes of *Homo* and *Australopithecus*



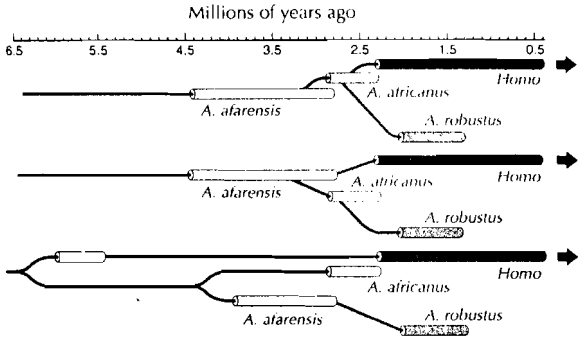
TRENDS IN HOMINID FOSSILS

Many hominid fossils have been found, dated and assigned to a species. These fossils show evolutionary trends, including increasing adaptation to bipedalism and increasing brain size. Other trends and dates of emergence are shown in the figure (right). *Australopithecus and Homo habilis* fossils were all found in Southern or Eastern Africa. *Homo erectus* fossils were found in Eastern Africa, but also in Asia, indicating that there was migration out of Africa. *Homo neanderthalensis* fossils were found in Europe and *Homo sapiens* in many parts of the world indicating further migrations.

EVOLUTIONARY RELATIONSHIPS BETWEEN HOMINIDS

There are many gaps in the hominid fossil record and so it is far from clear how species of hominid evolved. Three hypotheses for the origin of *Homo* are shown below.

Hypotheses for human origins



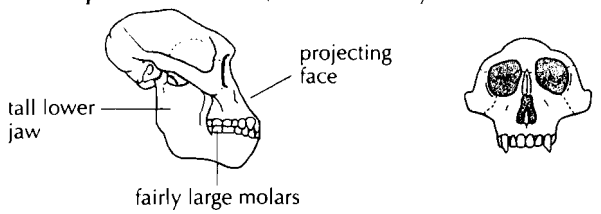
ECOLOGICAL CHANGES AND HOMINID ORIGINS

Five million years ago Africa became drier. Dense forest was replaced by thinner woodland with clearings. This may have prompted the evolution of bipedalism, although the earliest hominids probably still lived partly in trees. The powerful jaws and teeth of *Australopithecus* indicate a mainly vegetarian diet. About 2.5 million years ago Africa became much cooler and drier. Savannah grassland replaced forest. This change of habitat may have prompted the evolution of the first species of *Homo*, with the development of increasingly sophisticated tools and a change to a diet that included meat obtained by hunting and killing large animals. *Homo erectus* and later species developed the use of fire and were able to colonise colder areas and survive during ice ages.

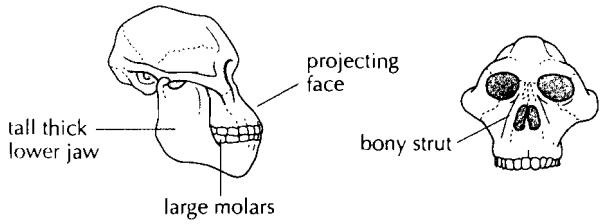
CULTURAL EVOLUTION

The large brains of *Homo sapiens* and other species of *Homo* allow much to be learned, both during the long period of childhood and during adulthood. Language, tool-making skills, hunting techniques, methods of agriculture, religion, art and many other forms of behaviour are passed on from one generation of a tribe or other group to the next by teaching and learning. These things are the culture of the group. New methods, inventions or customs can be incorporated into what is passed on. This is called cultural evolution. Cultural evolution has been very important in the recent evolution of humans and has allowed much more rapid change than genetic evolution could alone.

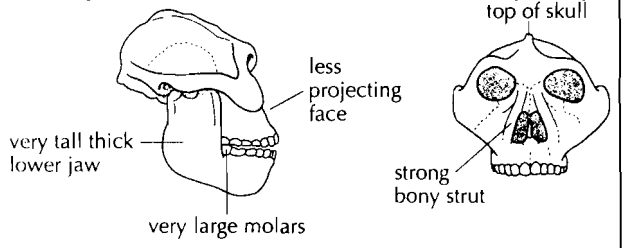
Australopithecus afarensis (4 to 2.5 million years)



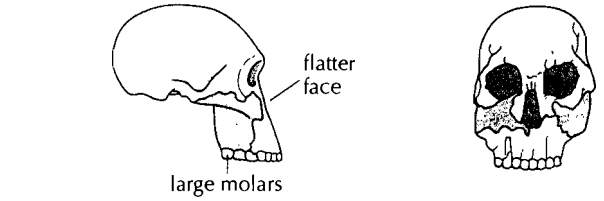
Australopithecus africanus (3 to <2.5 million years)



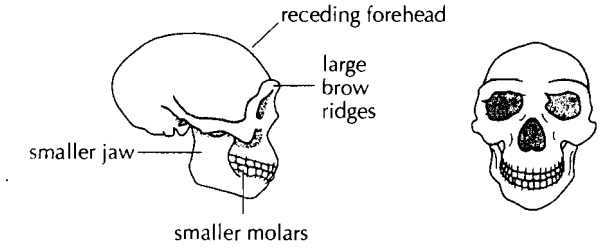
Australopithecus robustus (2 to 1 million years)



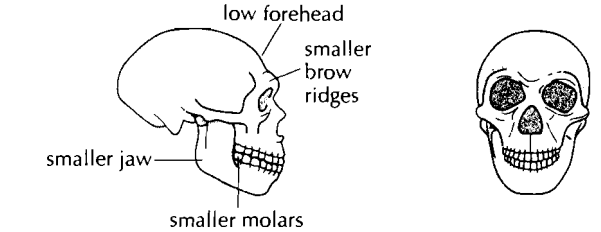
Homo habilis (2.4 to 1.6 million years)



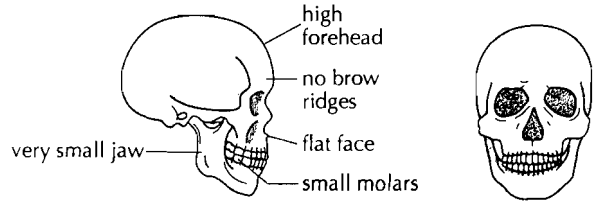
Homo erectus (1.7 to 1.8 million years)



Homo neanderthalensis (500000 years)



Homo sapiens (140000 to 70000 years)



HL Population genetics

THE HARDY–WEINBERG PRINCIPLE

If there are two alleles of a gene in a population, there are three possible genotypes – homozygous for each of the two alleles and heterozygous. The frequency of the two alleles in the population is usually represented by the letters *p* and *q*. The total frequency of the alleles in the population is 1.0, so

$$p + q = 1.$$

If there is random mating in a population, the chance of inheriting two copies of the first of the two alleles is $p \times p$. The chance of inheriting two copies of the second of the two alleles is $q \times q$. The expected frequency of the two homozygous genotypes is therefore p^2 and q^2. The expected frequency of the heterozygous genotype is $2pq$. The sum of all of these frequencies is 1.

$$p^2 + 2pq + q^2 = 1$$

This is called the **Hardy–Weinberg equation**. If the allele frequencies and the genotype frequencies in a population are known, this equation can be tested. The example below shows the results of a survey of the MN blood group gene in Japanese town. The two alleles of this gene are codominant.

Allele frequencies in the parental generation

M allele: $p = 0.525$ N allele: $q = 0.475$

Genotype frequencies in the offspring

	Predicted	Actual
MM	$p^2 = 0.276$	0.274
MN	$2pq = 0.499$	0.502
NN	$q^2 = 0.225$	0.224

The results of the survey show that the actual genotypes fit those predicted by the Hardy–Weinberg equation very closely. They therefore follow the **Hardy–Weinberg Principle**. Various conditions can prevent the Hardy–Weinberg Principle from operating – non-random mating, natural selection, mutation, small population size, immigration or emigration.

CALCULATING ALLELE AND GENOTYPE FREQUENCIES USING THE HARDY–WEINBERG EQUATION

If a population is known to be following the Hardy–Weinberg Principle, the Hardy–Weinberg equation can be used to calculate unknown frequencies. An example of this is a gene with two alleles that controls the ability to taste phenylthiocarbamide (PTC). The ability to taste PTC is due to the dominant allele (*T*) and non-tasting is due to the recessive allele (*t*).

1600 people were tested in a survey. 461 were non-tasters – a frequency of 0.288. Their genotype was homozygous recessive (*t t*).

If q = frequency of *t* allele, $q^2 = 0.288$ so $q = 0.537$

If p = frequency of *T* allele, $p = (1 - q) = 0.463$

The frequency of homozygous dominants (TT) and heterozygotes (Tt) can be calculated.

p^2 = frequency of homozygous dominants

$p^2 = 0.463 \times 0.463 = 0.214$

$2pq$ = frequency of heterozygotes

$2pq = 2\ (0.463 \times 0.537) = 0.497$

GENE POOLS

A new individual, produced by sexual reproduction inherits genes from its two parents. If there is random mating, any two individuals in an interbreeding population could be the two parents, so the individual could inherit any of the genes in the interbreeding population. These genes are called the **gene pool.**

A gene pool is all the genes in an interbreeding population.

NATURAL SELECTION AND CHANGES TO THE GENE POOL

If an allele increases the chances of survival and reproduction of individuals that possess it, the frequency of the allele in the gene pool will tend to increase. Conversely, if the allele reduces chances of survival and reproduction, it will decrease in frequency. These changes are due to natural selection.

The Hardy–Weinberg Principle can be used to test for natural selection. If allele and genotype frequencies in a population show that the Hardy–Weinberg Principle is being followed for a particular gene, this indicates that there is no natural selection. Members of the population all have an equal chance of survival whatever alleles of the gene they possess. The allele frequencies will not change between one generation and the next. If allele and genotype frequencies do not follow the Hardy–Weinberg Principle, a possible reason is that natural selection favours one allele over another.

Adaptations develop in populations as a result of changes in allele frequencies in the gene pool. This is sometimes called **microevolution**.

A population in which there are two alleles of a gene in the gene pool is **polymorphic**. If one allele is gradually replacing the other, the population shows **transient polymorphism**. Populations of ladybug that changed from having red wing cases with black spots to black wing cases are an example of transient polymorphism.

MACROEVOLUTION, GRADUALISM AND PUNCTUATED EQUILIBRIUM

Over long periods of time, many advantageous alleles will appear and spread through a species. These micro-evolutionary steps together constitute **macroevolution**. Eventually the amount of evolution becomes so great that the species is no longer the same – one species has evolved into another.

There has been much discussion among biologists about rates of evolution. One idea, called gradualism, is that evolution proceeds very slowly, but over long periods of time large changes can gradually take place. This does not fit in with the fossil record, which shows periods of stability, with fossils showing little evolution, followed by periods of sudden major change. The periods of stability may be due to equilibrium where living organisms have become well adapted to their environment so natural selection acts to maintain their characteristics. The periods of sudden change that punctuate the equilibrium may correspond with rapid environmental change, caused for example by volcanic eruptions or meteor impacts. New adaptations would be necessary to cope with new environmental conditions, hence strong directional selection and rapid evolution.

CHROMOSOME MUTATIONS

Mutations are changes to genes or chromosomes. Although mutations occur by chance, the rate at which they occur can be predicted. Down's syndrome and Klinefelter's syndrome are examples of conditions caused by chromosome mutations. Down's syndrome is described on page 21. Klinefelter's syndrome is caused by males having one or more extra X chromosomes (XXY). Although recognizably male, those with the syndrome have low testosterone levels and so are infertile and do not fully develop the male secondary sexual characteristics. Chromosome mutations often cause infertility and so the variation that they cause is not inherited. They are therefore not usually significant in evolution.

GENE MUTATIONS

Sickle cell anemia, cystic fibrosis and phenylketonuria (PKU) are diseases caused by gene mutations. PKU is caused by mutations of an autosomal gene that codes for phenylalanine hydroxylase. This enzyme converts the amino acid phenylalanine into tyrosine. Without it, phenylalanine accumulates in the blood to a harmful level that can cause mental retardation and death in young children. Over 30 different alleles cause PKU. Natural selection has kept them at low frequencies in the human population because, until screening and treatment for the disease recently became possible, children homozygous for PKU alleles died at an early age. In a similar way, natural selection will keep alleles that cause other genetic diseases at a low level in the human population.

CYSTIC FIBROSIS

Cystic fibrosis is the commonest genetic disease in Europe. It is caused by mutations of a gene coding for a chloride channel. This protein transports chloride ions across membranes in epithelium cells. Without the chloride channels in the plasma membrane, mucus secreted by epithelia becomes thick and sticky and tends to block airways of the breathing system, causing respiratory infections. Although various mutations of the chloride channel gene can cause cystic fibrosis, 70% of cases are due to one mutant allele, in which three bases coding for phenylalanine have been deleted. The frequency of this allele in Europe can be estimated using the Hardy–Weinberg equation.

Frequency of cystic fibrosis in Europe is one birth in 2500.

70% of these are due to the commonest allele,
i.e. one birth in 1750 = 0.00057

If the frequency of this allele is q,
$q^2 = 0.00057$ so $q = 0.023$

All the other alleles of the gene have a combined frequency of $(1 - 0.023) = 0.977$

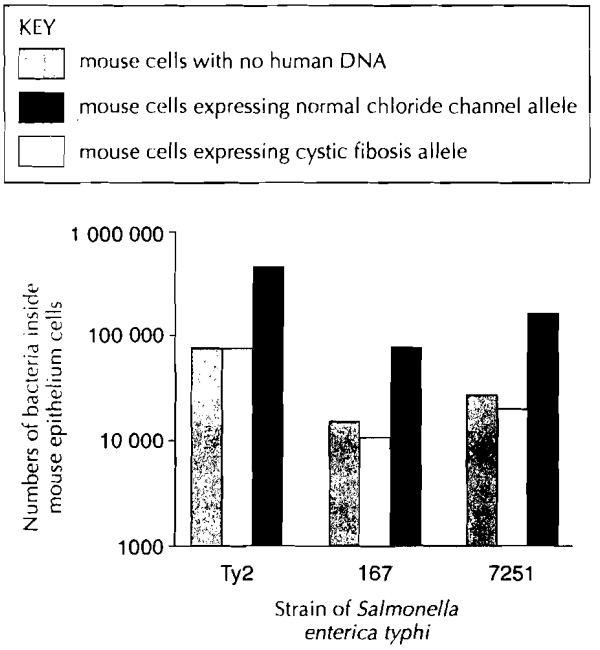
The estimated frequency of carriers of this allele can be calculated.
$2pq = 2(0.977 \times 0.023) = 0.045$

So, in Europe about 1in 20 people are carriers.

BALANCED POLYMORPHISMS

The allele that causes 70% of cystic fibrosis cases probably originated in one person in Europe about 50 000 years ago. Far from being eliminated by natural selection, it increased in frequency, despite causing a severe disease in homozygous individuals. The figure (below) shows the results of an experiment that shows a possible cause. The experiment involved genetically engineered mouse cells that expressed the cystic fibrosis allele. These cells and two groups of control cells were mixed with strains of *Salmonella enterica typhi*, the bacterium that cause typhoid fever.

Effect of human alleles on infection rates in mice



The results show that, in cells where the cystic fibrosis allele is expressed, S. *typhi* infection rates were much lower. Typhoid fever can cause death, so in populations where it is found, the cystic fibrosis allele might increase in frequency. Heterozygotes are the best adapted, because they are resistant to typhoid fever, but are do not develop cystic fibrosis. Natural selection will tend to maintain both the cystic fibrosis allele and the normal allele in the gene pool. It is not therefore a transient polymorphism and instead is called **balanced polymorphism**.

There is another well-known example of balanced polymorphism – sickle cell anemia (see page 28). Heterozygous individuals (Hb^AHb^S) do not develop sickle cell anemia and are resistant to malaria. They are therefore the best adapted in areas where malaria is found. The sickle cell allele has increased in frequency to high levels in some areas. In parts of Africa, as many as 40% of the population are carriers of the sickle cell allele, so show resistance to malaria. The Hardy – Weinberg equation can be used to calculate the frequency of the allele.

The frequency of carriers is 0.4
If the frequency of Hb^A is p
and the frequency of Hb^S is q,
$2pq = 0.4$.
So, $p = 0.724$ and $q = 0.276$

Species and speciation

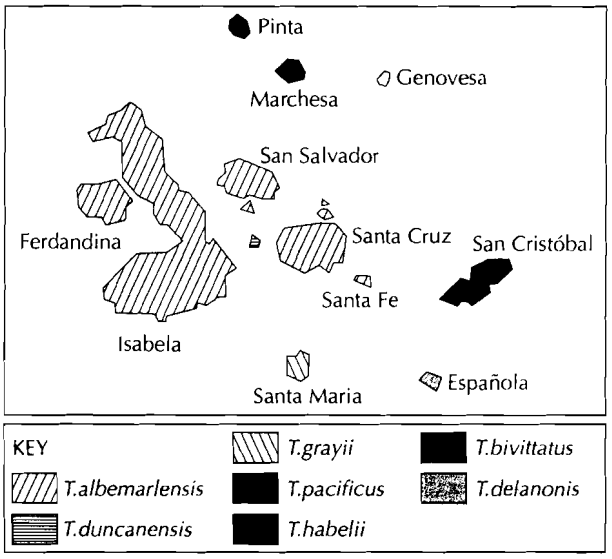
SPECIATION

The formation of new species is called speciation. New species are formed when a pre-existing species splits. This usually involves the isolation of a population from the remainder of its species and thus the isolation of its gene pool. The isolated population will gradually diverge from the rest of the species if natural selection acts differently on it. Eventually the isolated population will be unable to interbreed with the rest of the species – it has become a new species.

The most obvious way in which isolation can occur is by migration of members of a species to a new area that is geographically separated from the original territory of the species. This explains why there are often many **endemic species** (species that are found nowhere else) on isolated islands. The Galapagos finches are a well-known example.

The figure (below) shows the distribution of another type of animal on the Galapagos archipelago – lava lizards. The seven species are all endemic, although there are related species on the mainland of South America. They are present on twelve of the islands, but on each of these islands only one species is found. Six species are only found on one island. The other species is found on six islands, but there are behavioural differences between these six populations, so they have already begun to diverge.

Distribution of lava lizards on the Galapagos Islands



A second possible type of isolation is ecological. If two populations live in the same geographical area, but in different habitats, they may rarely interbreed. The apple maggot fly (*Rhagoletis pomonella*) of North America is an example of this. It originally only laid its eggs on hawthorn fruits, which were the food of its larvae. In the nineteenth century it started to infest non-native apple trees as well. Some strains of this species now prefer to lay their eggs on apple fruit and other strains prefer hawthorn fruit. Because the fruits ripen at different times, adults of the two strains emerge and mate at different times. They also inherit the preference either for apple or hawthorn and tend to remain on their preferred species. In addition to these behavioural differences, clear differences in allele frequencies have been found. If differences continued to build up, the two strains of *Rhagoletis pomonella* would eventually become separate species as a result of ecological isolation.

WHAT IS A SPECIES?

Biologists have been arguing about the exact meaning of the term species for over 200 years. Before the discovery that species can evolve, a species was regarded as a type of living organism with fixed characteristics, which distinguish it from other species. This is known as the morphological definition of a species. It is still a useful idea. Species can usually be distinguished from each other by their characteristics – this is how specimens are identified.

However the morphological definition does not recognize the fact that species evolve. If two populations with similar but not identical characteristics are geographically separated, they may be in the gradual process of splitting from one species into two separate ones. It is not easy for a taxonomist to decide whether to classify them as one or two species and some criterion is needed to decide. The reason for members of a species having common features is that they interbreed with each other. The reason for the characteristics of one species being different from those of another is that the two species do not interbreed and are evolving separately.

Biologists now regard interbreeding as a more important criterion than morphology. The biological definition of a species is *a group of actually or potentially interbreeding populations, with a common gene pool, which are reproductively isolated from other such groups.*

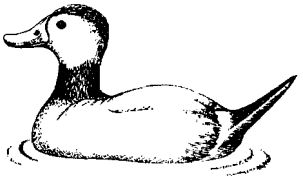
Only if two separated populations can be shown to be capable of interbreeding should they be classified as one species.

The biological species definition is widely accepted, but it does cause some problems.

- Many sibling species have been found. These are species that cannot interbreed, but show no significant differences in appearance. Although separate species, they are very difficult for ecologists to identify. For example, the Pipistrelle bat in Britain was recently shown to be two sibling species.
- Some pairs of species that are clearly different in their characteristics will interbreed. Many plant species can hybridize and some animals also can, including ruddy ducks and white-headed ducks (below).
- Some species always reproduce asexually, so the members of a population do not interbreed. The biological species definition is therefore unusable.
- Some species have spread around the Earth to form a series of interlinked but slightly different populations. If the ends of this series overlap, the populations are sometimes so different that they do not interbreed and behave as separate species. An example is herring gulls and lesser black backed gulls in north-west Europe, which are linked by a ring of populations around the northern hemisphere. Examples like this are called ring species.
- Fossils cannot be classified according to the biological species definition, as it is impossible to decide with which organisms they would have been able to interbreed.

Two animal species that can interbreed

White-headed duck



Ruddy duck