DNA, bones and human evolution

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Humans are perhaps the most successful of all modern living organisms, the only mammal to be found on every continent of the Earth. We are remarkably similar biologically to our nearest relatives but our cultures, communication skills and use of world resources make us appear very different from the rest of the animal kingdom. Yet in evolutionary terms we are a very recent addition to life on Earth and our understanding of the process of human evolution is still developing.

People are very excited about our growing understanding of the human genome and how the information in the DNA is interpreted to form the phenotype (the behaviour or characteristic determined by a gene) of the individual because this could have huge benefits for human health. These could range from predicting which embryos will result in healthy babies to developing personalised medicines which work as well as possible for each individual. This all involves looking to the future. Our increasing ability to sequence DNA and compare genomes is also proving very useful when it comes to analysing the past, and looking at how humans have evolved away from our closest relatives, the great apes.

Human relations

Humans (*Homo sapiens)* are mammals. More precisely, we are primates, a distinctive group of animals which first appeared more than 60 million years ago and which includes the lemurs, monkeys and apes. People are much more closely related to apes than to lemurs and monkeys, but all primates possess common features which we can recognise in ourselves. Many of the adaptations of primates are for an arboreal (tree-based) way of life. In people these basic evolutionary features have become further adapted to fit increasing numbers of niches.

Apes are our closest primate relatives - the skeletons of humans and African apes have every single bone in common, although the shape and size of some of them is very different. There are also strong genetic similarities between humans and apes.

Although humans only have 46 chromosomes whilst apes have 48, many of the individual chromosomes are remarkably similar. It has been shown that the amino acid sequences of human proteins are more than 99% identical to the amino acid sequences of the corresponding proteins in chimpanzees and gorillas.

Figure 1: Manipulative hands, large forward looking eyes, relatively unspecialised teeth and guts, long-term parental care of the offspring and larger than average brains are all typical features of primates.

Although we are biologically closely related to primates, modern humans are also very different in some striking and dramatic ways. Humans walk upright, and have a skeleton which is relatively light and well adapted for this style of locomotion. We are relatively hairless. We have a greatly expanded brain which enables us to develop and use tools of quite amazing complexity. The size and structure of the human brain enables us to construct and use language not merely to communicate simple biological drives such as sexual receptiveness, hunger and aggression but also complex ideas, plans and philosophies. The spurt of development seen during puberty in young humans is not seen in other primates. We have reduced brow ridges and a protruding chin. And finally human culture and the use of rituals to mark the various stages of life is unique in the animal kingdom.

How and when did the changes take place which moved us away from our common ancestors? No-one knows for certain - the models of human evolution are constantly changing, and there is still room for widely differing ideas because the evidence we have is open to different interpretations. The sort of evidence we use to build up a picture of human evolution includes fossils - unfortunately increasingly rare as we go backwards in time - comparative anatomy and physiology, archaeology (the study of a previous culture by excavation and examination of artefacts) and anthropology (the scientific study of human origins). But the study of DNA and the analysis of the genome is becoming more and more important in helping to build up a picture of how modern humans have evolved.

Common Ancestors

Ever since Charles Darwin first suggested that humans might share a common ancestor with the apes, people have wondered what that common ancestor might have been. However no single species which fills the role of the elusive ‘missing link’ between apes and humans has yet been found.

It seems unlikely that this missing link could be identified as the subtle differences which occurred around 6 million years ago and gave rise to ancestral apes and ancestral hominids, were so minute.

One way to begin building up a picture of the evolution of primates is by looking at the biochemistry of the primate group. This gives us a kind of molecular clock for when the different primate groups split and then evolved in different directions. The method is based on the immunological distance between species, and involves a close study of the DNA of the members of the primate group. Gorillas, chimps and human beings have more than 99% of their DNA in common, we are more closely related to chimps than we are to gorillas.

Once the hominid line had split from that of the other primates, a wide range of different hominids evolved. All but one of the known hominid species became extinct, but they all play a role in the story of human evolution. So who and what were these early hominids, and how do we know anything about them?

Determining the sequence of human evolution is fraught with difficulties due to the lack of evidence. The search for our ancestors begins with the evolution of the primates, but the primate fossils which we need as evidence are rare. This is partly because early primates, like most modern ones, lived in forest environments which have only occasionally been preserved as part of the fossil record. In addition, primate numbers have never been particularly high, and their bones are less robust than many other species making them less likely to become fossilised. This combination of factors leaves us only tantalising glimpses of primate evolution, far from the detailed picture we would ideally like.

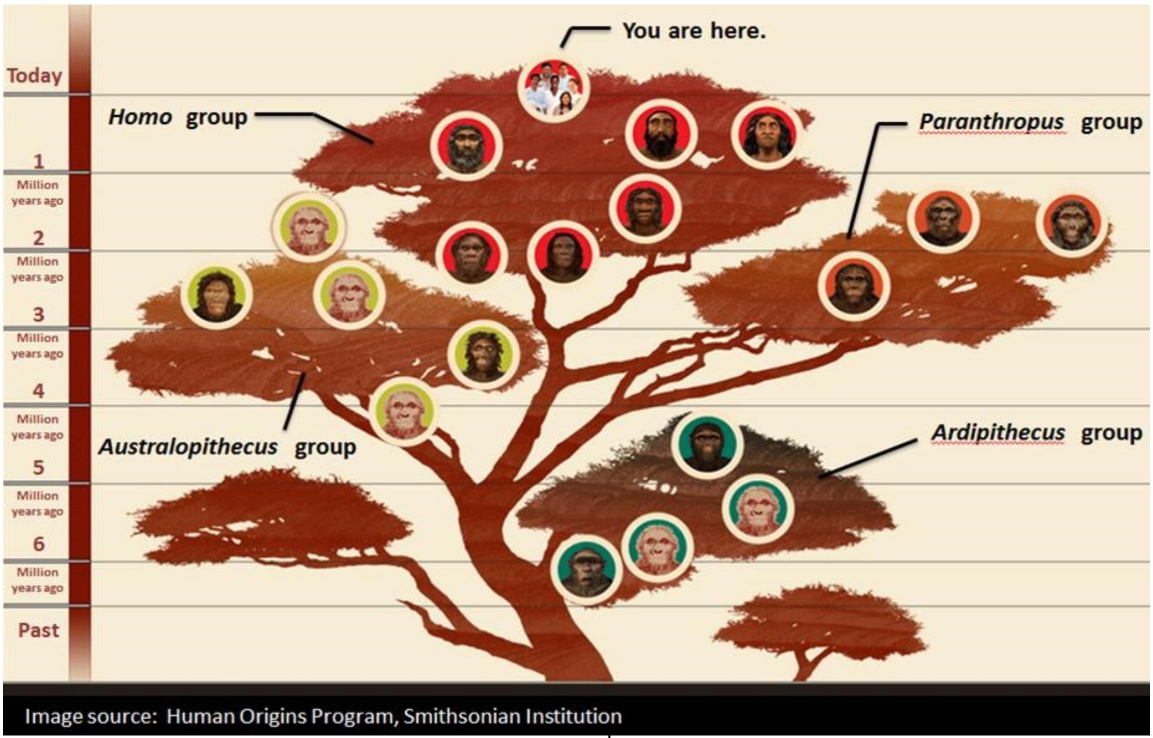

Figure 2: Our current picture of the evolution of hominids can be summarised diagrammatically, positioning the early hominids both relative to each other and with respect to the real passage of time. Image courtesy of the Smithsonian Museum.

Interpreting the evidence of the bones

Because evidence for hominid evolution is sparse it must be interpreted as widely as possible in order to maximise the information from it. For example, a fossil jaw and teeth can give an idea of the lower face of an individual, the shape of the jaw and the number of teeth. The shape, size and wear on the teeth along with any signs of damage and decay can also be used to deduce the sort of diet eaten and the care taken with the teeth. Similarly a pelvis or limb bone reveals evidence of the size and height of an individual, but can also be used to make deductions about posture and the degree of upright walking.

Because much of the evidence we have is very limited, a small new piece of evidence can have a big effect on our models.

For example, it was thought for many years that the early hominids could not walk upright properly. However a complete knee joint found at Hadar in 1973 by Taieb and Johanson moved those ideas backwards by about a million years.

From the evidence of this knee joint we learnt that hominids walked upright as their normal way of getting about up to 3 million years ago. Anthropologists must take the evidence available and use it to build up as clear a picture as they can of our earlier ancestors. Professor Alice Roberts examined the shoulder joints of modern humans, early humans and apes looking for arthritic shoulder disease from an evolutionary point of view to see if there were links with anatomical changes. She also worked with teams of other specialist scientists to make a TV series looking at how human ancestors evolved and some of the key anatomical changes which led to the success of modern humans. Pictured here is Alice Roberts with the skull of *Homo floresiensis ,* nicknamed the 'Hobbit', this was a rare find of a complete skull discovered in 2003 on the island of Flores in Indonesia. The remains

have been the subject of much research to determine whether they represent a species distinct from modern humans.

The genome speaks

The analysis of DNA from our early ancestors had other secrets to reveal. For many years it was accepted by most scientists that as the ancestors of modern humans spread out from Africa they replaced other early hominids such as *Homo neanderthalensis* (Neanderthal man).

Using fossil evidence, anthropologists have built up a picture of what early hominid species may have looked like. The evidence is in some cases scant, often no more than a fossilised tooth or jaw.

The genome speaks

No-one can be certain of when and where the hominids evolved away from the apes and so when *Homo sapiens* began their recognisable occupation of the Earth. But most of the reliable evidence points towards the continent of Africa as the site of much early evolution. In Africa we find our closest living relatives, a variety of fossil hominid remains, fossil hominid footprints, stone tools and evidence of the use of fire. In fact, *H. erectus* (see fig 2) appears to have left Africa and begun the great migration of people across the other continents of the world only in the last million or so years. Climatic changes and geological events - such as the creation of rift valleys - created the isolated environments and groups of individuals which drove evolutionary change through actual or effective reproductive isolation.

Another more recent idea is that all living human populations are derived from a single woman living in Africa around 200,000 years ago - the so called ‘African Eve’. Evidence for this idea comes from analysis of a very particular kind of genetic material – our mitochondrial DNA. Mitochondrial DNA is inherited only from our mothers, through the mitochondria present in the ovum. Each woman inherited her mitochondrial DNA from her mother and so on back in time. Each son is a dead end for the mitochondrial DNA, which is only passed on to the next generation through daughters. Mitochondrial DNA does mutate from time to time, but there is less variety in it than might be expected. This is taken to suggest that perhaps as recently as 200,000 years ago, perhaps as long ago as 500,000 years ago, there was a common female ancestor of all living people.

The recent discovery of the 4,000 year old remains of a hominid in the Cheddar Gorge raised particular excitement when mitochondrial DNA testing showed that ‘Cheddar Man’ had living relatives.

Adrian Targett lives near Cheddar Gorge and has mitochondrial DNA which matches that of the ancient hominid so closely that it is almost certain they shared a common female ancestor - ‘Cheddar Eve’ perhaps. It is an ironic twist that as this particular descendent is male with no siblings, it is the end of the line for her mitochondrial DNA. Craig Dent from Australia and two local children have also been found to have similar mitochondrial DNA. This ancient DNA line continues in many different descendants, and the story may add strength to the arguments for an African Eve on a larger scale.

The DNA revolution

2013 is a great year for deoxyribonucleic acid, or DNA as most of us know it. 60 years ago in 1953, James Watson and Francis Crick developed the now world famous double helix structural model for DNA, using the work of Maurice Wilkins and Rosalind Franklin to help them. For the first time scientists understood how DNA acted as the material of inheritance.

50 years later - only 10 years ago - another massive breakthrough came along. This was the completion of the Human Genome Project, which set out to identify the 20-25,000 human genes and the sequences of around 3 billion base pairs that make up human DNA. Since this amazing achievement, based on the collaborative work of 20 different research centres in 13 different countries, work on the human genome has gathered pace. Our knowledge and understanding moves forward all the time.

Figure 3: Our understanding of the role of DNA in genetics and the human genome has changed tremendously in the last 60 years.

Mixing the genes

The analysis of DNA from our early ancestors had other secrets to reveal. For many years it was accepted by most scientists that as the ancestors of modern humans spread out from Africa they replaced other early hominids such as *Homo neanderthalensis* (Neanderthal man). However in 2010 scientists succeeded in extracting and revealing about 60% of the Neanderthal genome. They then compared this with the genomes of modern humans from several different continents. Amazingly, people in Europe, Asia and New Guinea share up to 2.5% of their DNA with Neanderthals

Also in 2010, remains from a previously unknown offshoot of the Neanderthals were found in Siberia. The DNA was analysed and it was discovered that people in Melanesia and native Australians share up to 5% of their DNA with this Neanderthal group.

The first reaction of the scientific community was that this provided clear evidence of unexpected interbreeding between Neanderthals and modern human ancestors. Now scientists are divided. Some are still convinced that interbreeding between the two groups took place. Others have recently published evidence which pours cold water on these ideas. New dating techniques used on the fossil evidence appear to show that the two different groups of hominids did not live at the same time in the same area. This has led to suggestions that the shared DNA comes through from a shared common ancestor between Neanderthals and modern humans. This debate is still very much alive!

As more remains are discovered, and the use of DNA and dating technology continues to develop, the details of the story of our human evolution will come into ever-clearer focus.

Further reading:

Happy Birthday DNA *The Biologist* April/May 2013

The bone collector *The Biologist* April/May 2013

Life, the universe and everything *The Biologist* Feb/Mar 2013

http://www.nhm.ac.uk/nature-online/life/human-origins/modern-human-evolution/where/index.html

http://humanorigins.si.edu/evidence/human-family-tree - website on human evolution developed by the Smithsonian Institution

http://www.nhm.ac.uk/about-us/news/2011/january/ancient-denisovans-and-the-human-family-tree93500.html

http://www.bbc.co.uk/programmes/b013gmh1/episodes/guide

http://humanorigins.si.edu/evidence/genetics/ancient-dna-and-neanderthals/interbreeding - on possible interbreeding between Neanderthals and modern humans

http://www.bbc.co.uk/news/science-environment-19250778 BBC piece on the new report from Cambridge which cast doubt on the interbreeding theory