The genetic legacy of the Neanderthals

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New research published in a recent issue of *Nature* presents the sequencing of the entire genome of a Neanderthal woman who lived in the Altai Mountains in southern Siberia over 50,000 years ago. The data gathered from this study, along with another published study regarding the genome sequencing of a Denisovan hominin, a related group of humans who lived side by side with Neanderthals during this period, has much to say about prehistoric human development and the genetic makeup of modern humans, going far beyond any previous genetic research on archaic humans.

Genome sequencing is a relatively new process that enables researchers to map and examine DNA. The most famous example has been the Human Genome Project, an international effort to map the entire genetic sequence of modern humans all over the world, allowing for a detailed exploration of our biological history. The recent sequencing of Neanderthal and Denisovan genomes are the very first performed on extinct hominins (humans) and dramatically aid in our understanding of gene flow and drift between our archaic predecessors.

The Neanderthal type specimen fossil was discovered in a limestone quarry in the Neander Valley near Düsseldorf, Germany, in August 1856. Its discovery took place just three years prior to the publishing of Charles Darwin's *On the Origin of Species*, launching the finding into a larger debate about biological evolution. The most commonly debated aspect of Neanderthal history in recent years has been whether Neanderthals interbred with Homo sapiens and whether such offspring were fertile.

There are currently known remains attributed to 400 different Neanderthals ranging geographically from Portugal to Siberia, including northern European finds in England and Germany as well as Middle Eastern counterparts in Israel and Iraq. These fossils range in date from 350,000 years ago to 35,000 years ago.

Earlier Neanderthal fossils share traits with the older hominin Homo heidelbergensis, which spread across Africa, Europe, and Asia at least 600,000 years ago. Both Neanderthals and Homo sapiens are believed to have evolved from Homo heidelbergensis, each group developing in isolation due to prolonged quaternary glacial periods. If this hypothesis is correct, Homo sapiens would have developed in Africa, while Neanderthals evolved throughout Eurasia.

Neanderthals share 99.7 percent of their DNA with modern humans but display very specific morphological differences. Neanderthals were considerably more "robust" than Homo sapiens, featuring thicker bones and a larger brain case. While modern humans have an average brain capacity of 1400cc, the average Neanderthal reached 1600cc in size. Neanderthals are also thought to have been much stronger than anatomically modern humans, especially having stronger arms and hands. It has even been suggested that the more robust Neanderthal teeth were used as cutting tools (the more "gracile" Homo sapiens teeth perhaps developed along with the increased use of fire to cook food, which made it softer). Neanderthals are also thought to have consumed a larger percentage of meat as part of their diet, including big game animals. One of the more popular explanations for the physical divergence between Neanderthals and Homo sapiens is climatic. Bodies with shorter limbs and thicker forms retain heat more efficiently in colder geographic areas while more lissome figures with longer limbs disperse heat better in warmer climates.

In 2008, an excavation performed in Denisova Cave in southern Siberia, near the borders of Kazakhstan and Mongolia, yielded the fossil remains of both Neanderthals and those of a potentially new group of hominins that have since become known as "Denisovans." These remains have enabled researchers to determine that both groups inhabited Eurasia during the period when modern humans emerged from Africa (around 50,000 years ago), leading some to believe that Denisovans similarly evolved from Homo heidelbergensis living in Asia.

According to the recent *Nature* article, the "Altai Neanderthal" genome was sequenced from DNA extracted from a toe phalanx discovered in Denisova Cave in 2010. The archaeological layer within which the toe was found is located in the east gallery of the cave and is thought to be at least 50,000 years old. While the bone features traits commonly found in both Neanderthals and modern humans,

the DNA found within shares a common ancestor with six previously published Neanderthal DNA sequences, providing adequate evidence that it indeed belongs to a Neanderthal.

The main finding of the report, which has been focused on by most media outlets, is a high level of inbreeding in the Altai Neanderthal. According to the published research, her parents were closely related enough to be either half-siblings who shared the same mother or of another close relation such as double first cousins, uncle and niece, or grandmother and grandson. While there has been a high level of discussion about whether inbreeding was common amongst the Altai Neanderthal population, no scientific evidence has surfaced which supports this theory. Inbreeding is more typical of small, isolated populations.

The toe phalanx genome was compared to several other hominin genomes, including the Denisovan genome sequence that was previously recorded from DNA extracted from a finger phalanx. The Denisovan finger, excavated in 2008, was found within the same archaeological layer as the Neanderthal toe. Other genomes used for comparison include those of several other Neanderthals and 25 present-day humans. Researchers were thus able to provide new estimates of population split times.

Their estimates suggest that the ancestors of modern humans split from both Neanderthals and Denisovans between approximately 553,000 and 589,000 years ago, while the Neanderthal and Denisovan populations seem to have split apart later, about 381,000 years ago.

According to the authors of the *Nature* article and of potentially greater interest is the fact that the population of the ancestors of modern humans began to increase over time after the split, while Neanderthals and Denisovans saw subsequent decreases in population size. This supports previous speculations that Neanderthal populations were small and stable or even declining at the height of the last glacial period.

The new genome sequences also support admixture theories previously suggested by researchers. Much controversy has surrounded the suggestion that archaic humans such as Neanderthals and Denisovans interbred with the ancestors of modern humans, especially amongst those who theorize that other hominin groups were violently eradicated by “blood-thirsty” Homo sapiens. Other earlier writers on the subject placed Neanderthals as direct ancestors of modern humans while more recent scholarly work has suggested a smaller percentage of Neanderthal genetic material in the Homo sapiens genome sequence.

In recent years, new research has strongly indicated that modern humans emerged and dispersed from Africa approximately 50,000 years ago, mixing with Neanderthals in the Middle East before venturing into Europe and Asia, with those anatomically-modern humans who passed into Asia mixing further with Denisovans in Oceania. If these archaic groups intermingled genetically, a newer, clearer picture of our ancestors can be understood, a picture that is far less grim than the violent images of conquest propounded by some analysts.

The Altai Neanderthal genome sequence shows that Neanderthal-derived DNA in all non-Africans is 1.5 to 2.1 percent, while Denisovan-derived DNA found in human inhabitants of Papua New Guinea and Australia is 3 to 6 percent. There are also small traces (0.5 percent) of Denisovan DNA in mainland Asian and Native American populations. Such evidence of archaic human gene flow into modern populations suggests that decreasing populations of Neanderthals and perhaps also Denisovans never became “extinct” but were merely subsumed by increasing numbers of modern humans.

Ultimately this data supports the theory that Neanderthals and Denisovans did interbreed with Homo sapiens and did produce fertile offspring.

Absolutely integral to an understanding of archaic humanity is consciousness of the fact that very much is still unknown to us. In fact, if all of the excavated hominin remains from which we derive our knowledge of our ancestors were gathered together into one collection, it would easily fit into the bed of a single pickup truck.

Several other lineages and archaic human populations are for now beyond our knowledge. For example, 2.7 to 5.8 percent of the Denisovan genome comes from an unknown source, possibly an archaic hominin group which diverged from other hominins around 1 million years ago. Therefore, our current understanding of archaic humans must remain pliable and open to new findings and interpretations of raw data.