What does it mean to be human?

March 7 2017, by Gaia Vince

The Rock of Gibraltar appears out of the plane window as an immense limestone monolith sharply rearing up from the base of Spain into the Mediterranean. One of the ancient Pillars of Hercules, it marked the end of the Earth in classical times. Greek sailors didn't go past it. Atlantis, the unknown, lay beyond.

In summer 2016, Gibraltar is in the throes of a 21st-century identity crisis: geographically a part of Spain, politically a part of Britain; now torn, post Brexit, between its colonial and European Union ties. For such a small area – less than seven square kilometres – Gibraltar is home to an extraordinarily diverse human population. It has been home to people of all types over the millennia, including early Europeans at the edge of their world, Phoenicians seeking spiritual support before venturing into the Atlantic, and Carthaginians arriving in a new world from Africa.

But I've come to see who was living here even further back, between 30,000 and 40,000 years ago, when sea levels were much lower and the climate was swinging in and out of ice ages. It was a tough time to be alive and the period saw the species that could, such as birds, migrate south to warmer climes, amid plenty of local extinctions. Among the large mammal species struggling to survive were lions, wolves and at least two types of human: our own 'modern human' ancestors, and the last remaining populations of our cousins, the Neanderthals.

By understanding more about these prehistoric people, we can learn about who we are as a species today. Our ancestors' experiences shaped us, and they may still hold answers to some of our current health problems, from diabetes to depression.

Everyone of European descent has some Neanderthal DNA in their genetic makeup

I'm picked up outside my hotel by archaeologists Clive and Geraldine Finlayson, in a car that itself looks fairly ancient. Typical for this crowded little peninsula, they are of diverse origins – he, pale-skinned and sandy-haired, can trace his ancestry back to Scotland; she, olive-skinned and dark-haired, from the Genoese refugees escaping Napoleon's purges. How different we humans can look from each other. And yet the people whose home I am about to visit truly were of a different race.

We don't know how many species of humans there have been, how many different races of people, but the evidence suggests that around 600,000 years ago one species emerged in Africa that used fire, made simple tools from stones and animal bones, and hunted big animals in large cooperative groups. And 500,000 years ago, these humans, known as Homo heidelbergensis, began to take advantage of fluctuating climate changes that regularly greened the African continent, and spread into Europe and beyond.

By 300,000 years ago, though, migration into Europe had stopped, perhaps because a severe ice age had created an impenetrable desert across the Sahara, sealing off the Africans from the other tribes. This geographic separation enabled genetic differences to evolve, eventually resulting in different races, although they were still the same species and would prove able to have fertile offspring together. The race left behind in Africa would become Homo sapiens sapiens, or 'modern humans'; those who evolved adaptations to the cooler European north would become Neanderthals, Denisovans and others whom we can now only glimpse with genetics.

Neanderthals were thriving from Siberia to southern Spain by the time a few families of modern humans made it out of Africa around 60,000 years ago. These Africans encountered Neanderthals and, on several occasions, had children with them. We know this because human DNA has been found in the genomes of Neanderthals, and because everyone alive today of European descent – including me – has some Neanderthal DNA in their genetic makeup. Could it be that their genes, adapted to the northerly environment, provided a selective advantage to our ancestors as well?

It was like Neanderthal City

After driving through narrow tunnels on a road that skirts the cliff face, we pull up at a military checkpoint. Clive shows the guard our accreditation and we're waved through to park inside. Safety helmets on to protect from rockslides, we leave the car and continue on foot under a low rock arch. A series of metal steps leads steeply down the cliff to a narrow shingle beach, 60 metres below. The tide is lapping the pebbles and our feet must negotiate the unstable larger rocks to find a dry path.

I've been concentrating so hard on keeping my footing that it is something of a shock to look up and suddenly face a gaping absence in the rock wall. We have reached Gorham's Cave, a great teardrop-shaped cavern that disappears into the white cliff face and, upon entering, seems to grow in height and space. This vast, cathedral-like structure, with a roof that soars high into the interior, was used by Neanderthals for tens of thousands of years. Scientists believe it was their last refuge. When Neanderthals disappeared from here, some 32,000 years ago, we became the sole inheritors of our continent.

I pause, perched on a rock inside the entrance, in order to consider this –

people not so different from myself once sat here, facing the Mediterranean and Africa beyond. Before I arrived in Gibraltar, I used a commercial genome-testing service to analyse my ancestry. From the vial of saliva I sent them, they determined that 1 per cent of my DNA is Neanderthal. I don't know what health advantages or risks these genes have given me – testing companies are no longer allowed to provide this level of detail – but it is an extraordinary experience to be so close to the intelligent, resourceful people who bequeathed me some of their genes. Sitting in this ancient home, knowing none of them survived to today, is a poignant reminder of how vulnerable we are – it could so easily have been a Neanderthal woman sitting here wondering about her extinct human cousins.

Gorham's Cave seems an oddly inaccessible place for a home. But Clive, who has been meticulously exploring the cave for 25 years, explains that the view was very different back then. With the sea levels so much lower, vast hunting plains stretched far out to sea, letting people higher on the rock spot prey and signal to each other. In front of me would have been fields of grassy dunes and lakes – wetlands that were home to birds, grazing deer and other animals. Further around the peninsula to my right, where the dunes gave way to shoreline, would have been clam colonies and mounds of flint. It was idyllic, Clive says. The line of neighbouring caves here probably had the highest concentration of Neanderthals living anywhere on Earth. "It was like Neanderthal City," he adds.

Deep inside the cave, Clive's team of archaeologists have found the remains of fires. Further back are chambers where the inhabitants could have slept protected from hyenas, lions, leopards and other predators. "They ate shellfish, pine seeds, plants and olives. They hunted big game and also birds. There was plenty of fresh water from the springs that still exist under what is now seabed," Clive says. "They had spare time to sit and think – they weren't just surviving."

He and Geraldine have uncovered remarkable evidence of Neanderthal culture in the cave, including the first example of Neanderthal artwork. The 'hashtag', a deliberately carved rock engraving, is possibly evidence of the first steps towards writing. Other signs of symbolic or ritualistic behaviour, such as the indication that Neanderthals were making and wearing black feather capes or headdresses as well as warm clothes, all point to a social life not so different to the one our African ancestors were experiencing.

Clive shows me a variety of worked stones, bone and antler. I pick up a flint blade and hold it in my hand, marvelling at how the same technology is being passed between people biologically and culturally linked but separated by tens of thousands of years. Other sites in Europe have uncovered Neanderthal-made necklaces of strung eagle talons dating back 130,000 years, little ochre clamshell compacts presumably for adornment, and burial sites for their dead.

These people evolved outside of Africa but clearly had advanced culture and the capability to survive in a hostile environment. "Consider modern humans were in the Middle East perhaps 70,000 years ago, and reached Australia more than 50,000 years ago," says Clive. "Why did it take them so much longer to reach Europe? I think it was because Neanderthals were doing very well and keeping modern humans out."

But by 39,000 years ago, Neanderthals were struggling. Genetically they had low diversity because of inbreeding and they were reduced to very low numbers, partly because an extreme and rapid change of climate was pushing them out of many of their former habitats. A lot of the forested areas they depended on were disappearing and, while they were intelligent enough to adapt their tools and technology, their bodies were unable to adapt to the hunting techniques required for the new climate and landscapes.

"In parts of Europe, the landscape changed in a generation from thick forest to a plain without a single tree," Clive says. "Our ancestors, who were used to hunting in bigger groups on the plains, could adapt easily: instead of wildebeest they had reindeer, but effectively the way of capturing them was the same. But Neanderthals were forest people.

"It could've gone the other way – if instead the climate had got wetter and warmer, we might be Neanderthals today discussing the demise of modern humans."

Although the Neanderthals, like the Denisovans and other races we are yet to identify, died out, their genetic legacy lives on in people of European and Asian descent. Between 1 and 4 per cent of our DNA is of Neanderthal origins, but we don't all carry the same genes, so across the population around 20 per cent of the Neanderthal genome is still being passed on. That's an extraordinary amount, leading researchers to suspect that Neanderthal genes must be advantageous for survival in Europe.

Interbreeding across different races of human would have helped accelerate the accumulation of useful genes for the environment, a process that would have taken much longer to occur through evolution by natural selection. Neanderthal tweaks to our immune system, for example, may have boosted our survival in new lands, just as we prime our immune system with travel vaccines today. Many of the genes are associated with keratin, the protein in skin and hair, including some that are linked to corns and others that play a role in pigmentation – Neanderthals were redheads, apparently. Perhaps these visible variants were considered appealing by our ancestors and sexually selected for, or perhaps a tougher skin offered some advantage in the colder, darker European environment.

Some Neanderthal genes, however, appear to be a disadvantage, for instance making us more prone to diseases like Crohn's, urinary tract disorders and type 2 diabetes, and to depression. Others change the way we metabolise fats, risking obesity, or even make us more likely to become addicted to smoking. None of these genes are a direct cause of these complicated conditions, but they are contributory risk factors, so how did they survive selection for a thousand generations?

It's likely that for much of the time since our sexual encounters with Neanderthals, these genes were useful. When we lived as hunter-gatherers, for example, or early farmers, we would have faced times of near starvation interspersed with periods of gorging. Genes that now pose a risk of diabetes may have helped us to cope with starvation, but our new lifestyles of continual gorging on plentiful, high-calorie food now reveal harmful side-effects. Perhaps it is because of such latent disadvantages that Neanderthal DNA is very slowly now being deselected from the human genome.

While I can (sort of) blame my Neanderthal ancestry for everything from mood disorders to being greedy, another archaic human race passed on genes that help modern Melanesians, such as people in Papua New Guinea, survive different conditions. Around the time that the ancestors of modern Europeans and Asians were getting friendly with Neanderthals, the ancestors of Melanesians were having sex with Denisovans, about whom we know very little. Their surviving genes, however, may help modern-day Melanesians to live at altitude by changing the way their bodies react to low levels of oxygen. Some geneticists suspect that other, yet-to-be-discovered archaic races may have influenced the genes of other human populations across the world.

Interbreeding with Neanderthals and other archaic humans certainly changed our genes, but the story doesn't end there.

I am a Londoner, but I'm a little darker than many Englishwomen because my father is originally from Eastern Europe. We are attuned to such slight differences in skin colour, face shape, hair and a host of other less obvious features encountered across different parts of the world. However, there has been no interbreeding with other human races for at least 32,000 years. Even though I look very different from a Han Chinese or Bantu person, we are actually remarkably similar genetically. There is far less genetic difference between any two humans than there is between two chimpanzees, for example.

The reason for our similarity is the population bottlenecks we faced as a species, during which our numbers dropped as low as a few hundred families and we came close to extinction. As a result, we are too homogeneous to have separated into different races. Nevertheless, variety has emerged through populations being separated geographically – and culturally, in some cases – over thousands of years. The greatest distinctions occur in isolated populations where small genetic and cultural changes become exaggerated, and there have been many of them over the 50,000 years since my ancestors made the journey out of Africa towards Europe.

According to the analysis of my genome, my haplogroup is H4a. Haplogroups describe the mutations on our mitochondrial DNA, passed down through the maternal line, and can theoretically be used to trace a migratory path all the way back to Africa. H4a is a group shared by people in Europe, unsurprisingly, and western Asia. It is, the genome-testing company assures me, the same as Warren Buffet's. So what journey did my ancestors take that would result in these mutations and give me typically European features?

"I was dumped by helicopter in the wilderness with two other people, a Russian and an indigenous Yukaghir man, with our dogs, our guns, our traps, a little food and a little tea. There we had to survive and get food and furs in the coldest place on Earth where humans live naturally – minus 60 degrees."

Eske Willerslev lived for six months as a trapper in Siberia in his 20s. Separately, his identical twin brother Rane did the same. When they were teenagers, their father had regularly left them in Lapland to survive alone in the wilderness for a couple of weeks, fostering a passion for the remote tundra and the people who live there, and they went on increasingly lengthy expeditions. But surviving practically alone was very different. "It was a childhood dream, but it was the toughest thing I have ever done," Eske admits.

These experiences affected the twins deeply, and both have been driven towards a deeper understanding of how the challenge of survival has forged us as humans over the past 50,000 years. It led Eske into the science of genetics, and to pioneering the new field of ancient DNA sequencing. Now director of the Centre for GeoGenetics at the Natural History Museum of Denmark, Eske has sequenced the world's oldest genome (a 700,000-year-old horse) and was the first to sequence the genome of an ancient human, a 4,000-year-old Saqqaq man from Greenland. Since then, he has gone on to sequence yet more ancient humans and, in doing so, has fundamentally changed our understanding of early human migration through Europe and beyond. If anyone can unpick my origins, it is surely Eske.

First, though, I go to meet his twin Rane, who studied humanities, went into cultural anthropology and is now a professor at Aarhus University. He's not convinced that his brother's genetic approach can reveal all the answers to my questions: "There exists an uneasy relationship between biology and culture," he tells me. "Natural scientists claim they can reveal what sort of people moved around, and they are not interested in having their models challenged. But this cannot tell you anything about what people thought or what their culture was."

To put this point to Eske, I visit him in his delightful museum office, opposite a petite moated castle and in the grounds of the botanic gardens

– there could scarcely be a more idyllic place for a scientist to work. Greeting him for the first time, just hours after meeting Rane, is disconcerting. Identical twins are genetically and physically almost exactly the same – looking back, many years from now, at DNA left by the brothers, it would be all but impossible to tell them apart or even to realise that there were two of them.

Eske tells me that he is increasingly working with archaeologists to gain additional cultural perspective, but that genetic analysis can answer questions that nothing else can. "You find cultural objects in certain places and the fundamental question is: Does that mean people who made it were actually there or that it was traded? And, if you find very similar cultural objects, does that mean there was parallel or convergent cultural evolution in the two places, or does that mean there was contact?" he explains.

"For example, one theory says the very first people crossing into the Americas were not Native Americans but Europeans crossing the Atlantic, because the stone tools thousands of years ago in America are similar to stone tools in Europe at the same time. Only when we did the genetic testing could we see it was convergent evolution, because the guys carrying and using those tools have nothing to do with Europeans. They were Native Americans. So the genetics, in terms of migrations, is by far the most powerful tool we have available now to determine: was it people moving around or was it culture moving around? And this is really fundamental."

What Eske went on to discover about Native American origins rewrote our understanding completely. It had been thought that they were simply descendants of East Asians who had crossed the Bering Strait. In 2013, however, Eske sequenced the genome of a 24,000-year-old boy discovered in central Siberia, and found a missing link between ancient Europeans and East Asians, the descendants of whom would go on to populate America. Native Americans can thus trace their roots back to Europe as well as East Asia.

And what about my ancestors? I show Eske the H4a haplotype analysed by the sequencing company and tell him it means I'm European. He laughs derisively. "You could be and you could be from somewhere else," he says. "The problem with the gene-sequencing tests is that you can't look at a population and work back to see when mutation arose with much accuracy – the error bars are huge and it involves lots of assumptions about mutation rates.

"This is why ancient genetics and ancient genomics are so powerful – you can look at an individual and say, 'Now we know we are 5,000 years ago, how did it look? Did they have this gene or not?'"

The things that we thought we understood about Europeans are coming unstuck as we examine the genes of more ancient people. For example, it was generally accepted that pale skin evolved so we could get more vitamin D after moving north to where there was little sun and people had to cover up against the cold. But it turns out that it was the Yamnaya people from much further south, tall and brown-eyed, who brought pale skins to Europe. Northern Europeans before then were dark-skinned and got plenty of vitamin D from eating fish.

It is the same with lactose tolerance. Around 90 per cent of Europeans have a genetic mutation that allows them to digest milk into adulthood, and scientists had assumed that this gene evolved in farmers in northern Europe, giving them an additional food supply to help survive the long winters. But Eske's research using the genomes of hundreds of Bronze Age people, who lived after the advent of farming, has cast doubt on this theory too: "We found that the genetic trait was almost non-existent in the European population. This trait only became abundant in the northern European population within the last 2,000 years," he says.

It turns out that lactose tolerance genes were also introduced by the Yamnaya. "They had a slightly higher tolerance to milk than the European farmers and must have introduced it to the European gene pool. Maybe there was a disaster around 2,000 years ago that caused a population bottleneck and allowed the gene to take off. The Viking sagas talk about the sun becoming black – a major volcanic eruption – that could have caused a massive drop in population size, which could have been where some of that stock takes off with lactose."

While ancient genomics can help satisfy curiosity about our origins, its real value may be in trying to unpick some of the different health risks in different populations. Even when lifestyle and social factors are taken into account, some groups are at significantly higher risk of diseases such as diabetes or HIV, while other groups seem more resistant. Understanding why could help us prevent and treat these diseases more effectively.

It had been thought that resistance to infections like measles, influenza and so on arrived once we changed our culture and started farming, living in close proximity with other people and with animals. Farming started earlier in Europe, which was thought to be why we have disease resistance but Native Americans don't, and also why the genetic risks of diabetes and obesity are higher in native Australian and Chinese people than in Europeans.

"Then," says Eske, "we sequenced a hunter-gatherer from Spain, and he showed clear genetic resistance to a number of pathogens that he shouldn't have been exposed to." Clearly, Europeans and other groups have a resistance that other groups don't have, but is this really a result of the early agricultural revolution in Europe, or is something else going on?

Eske's analysis of people living 5,000 years ago has also revealed massive epidemics of plague in Europe and Central Asia, 3,000 years earlier than previously thought. Around 10 per cent of all skeletons the team analysed had evidence of plague. "Scandinavians and some northern Europeans have higher resistance to HIV than anywhere else in the world," Eske notes. "Our theory is that their HIV resistance is partly resistance towards plague."

It could be that the cultural changes we have made, such as farming and herding, have had less influence on our genes than we thought. Perhaps it is simply the randomness of genetic mutation that has instead changed our culture. There's no doubt that where mutations have occurred and spread through our population, they have influenced the way we look, our health risks and what we can eat. My ancestors clearly didn't stop evolving once they'd left Africa – we're still evolving now – and they have left an intriguing trail in our genes.

At the Gibraltar Museum, a pair of Dutch archaeology artists have created life-size replicas of a Neanderthal woman and her grandson, based on finds from nearby. They are naked but for a woven amulet and decorative feathers in their wild hair. The boy, aged about four, is embracing his grandmother, who stands confidently and at ease, smiling at the viewer. It's an unnerving, extraordinarily powerful connection with someone whose genes I may well share, and I recall Clive's words from when I asked him if modern humans had simply replaced Neanderthals because of our superior culture.

"That replacement theory is a kind of racism. It's a very colonialist mentality," he said. "You're talking almost as if they were another species."

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