Ancient human genome from southern Africa throws light on our origins

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Professor Vanessa Hayes in the field. Credit: Chris Bennett photography

What can DNA from the skeleton of a man who lived 2,330 years ago in the southernmost tip of Africa tell us about ourselves as humans? A great deal when his DNA profile is one of the 'earliest diverged' – oldest in genetic terms – found to-date in a region where modern humans are believed to have originated roughly 200,000 years ago.

The man's maternal DNA, or 'mitochondrial DNA', was sequenced to provide clues to early modern human prehistory and evolution. Mitochondrial DNA provided the first evidence that we all come from Africa, and helps us map a figurative genetic tree, all branches deriving from a common 'Mitochondrial Eve'.

When archaeologist Professor Andrew Smith from the University of Cape Town discovered the skeleton at St. Helena Bay in 2010, very close to the site where 117,000 year old human footprints had been found – dubbed "Eve's footprints" – he contacted Professor Vanessa Hayes, a world-renowned expert in African genomes.

At the time, Hayes was Professor of Genomic Medicine at the J. Craig Venter Institute in San Diego, California. She now heads the Laboratory for Human Comparative and Prostate Cancer Genomics at Sydney's Garvan Institute of Medical Research.

The complete 1.5 metre tall skeleton was examined by Professor Alan Morris, from the University of Cape Town. A biological anthropologist, Morris showed that the man was a 'marine forager'. A bony growth in his ear canal, known as 'surfer's ear', suggested that he spent some time diving for food in the cold coastal waters, while shells carbon-dated to the same period, and found near his grave, confirmed his seafood diet. Osteoarthritis and tooth wear placed him in his fifties.

Due to the acidity of the soil within the region, acquiring DNA from skeletons has proven problematic. The Hayes team therefore worked with the world's leading laboratory in ancient DNA research, namely that of paleogeneticist Professor Svante Pääbo at the Max Planck Institute for Evolutionary Anthropolgy in Leipzig, Germany, who successfully sequenced a Neanderthal.

The team generated a complete mitochondrial genome, using DNA

extracted from a tooth and a rib. The findings provided genomic evidence that this man, from a lineage now presumed extinct, as well as other indigenous coastal dwellers like him, were the most closely related to 'Mitochondrial Eve'.

The study underlines the significance of southern African archaeological remains in defining human origins, and is published in the journal *Genome Biology and Evolution*, now online.

"We were thrilled that archaeologist Andrew Smith understood the importance of not touching the skeleton when he found it, and so did not contaminate its DNA with modern human DNA," said Professor Hayes.

"I approached Svante Pääbo because his lab is the best in the world at DNA extraction from ancient bones. This skeleton was very precious and we needed to make sure the sample was in safe hands."

"Alan Morris undertook some incredible detective work. He used his skills in forensics and murder cases to assemble a profile of the man behind the St Helena skeleton."

"Alan helped establish that this man was a marine hunter-gatherer - in contrast to the contemporary inland hunter-gatherers from the Kalahari dessert. We were very curious to know how this man related to them."

"We also know that this man pre-dates migration into the region, which took place around 2,000 years ago when pastoralists made their way down the coast from Angola, bringing herds of sheep. We could demonstrate that our marine hunter-gatherer carried a different maternal lineage to these early migrants – containing a DNA variant that we have never seen before."

"Because of this, the study gives a baseline against which historic herders

at the Cape can now be compared."

While interested in African lineages, and how they interact with each other, Professor Hayes is especially keen for Africa to inform genomic research and medicine worldwide.

"One of the biggest issues at present is that no-one is assembling genomes from scratch – in other words, when someone is sequenced, their genome is not pieced together as is," she said.

"Instead, sections of the sequenced genome are mapped to a reference genome. Largely biased by European contribution, the current reference is poorly representative of indigenous peoples globally."

"If we want a good reference, we have to go back to our early human origins."

"None of us that walk on this planet now are pure anything - we are all mixtures. For example 1-4% of Eurasians even carry Neanderthal DNA"

"We need more genomes that don't have extensive admixture. In other words, we need to reduce the noise."

"In this study, I believe we may have found an individual from a lineage that broke off early in modern human evolution and remained geographically isolated. That would contribute significantly to refining the human reference genome."

More information: Paper - gbe.oxfordjournals.org/content …/gbe.evu202.full.pdf