

# Genetic analysis of ancient 'Iceman' mummy traces ancestry from Alps to Mediterranean isle

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A 5,300-year-old mummy, the basis for this reconstruction, has provided rare DNA samples that may shed light on ancient migration patterns. Credit: Reconstruction by Kennis, © South Tyrol Museum of Archaeology, photo by Ochsenreiter

The Iceman mummy, also known as Otzi, is about 5,300 years old. Scientists studying his body since his discovery in the Italian Alps in 1991 have learned many things, including the cause of his death (an arrow to the back) and his last meal (ibex meat). An analysis of the corpse's chemical composition suggested that he was born and lived his

entire life in the Tyrol area where his body was found. Now they're delving deeper to unearth more clues in the mummy's DNA.

On Feb. 28, European and American scientists, including Stanford School of Medicine genetics professor Carlos Bustamante, PhD, and senior research associate Peter Underhill, PhD, announced the sequencing of Otzi's entire genome. It is the oldest human sample to undergo such an analysis. Postdoctoral scholars Andres Moreno-Estrada, PhD; Brenna Henn, PhD; and Martin Sikora, PhD, also worked on the study, which appeared in *Nature Communications*. High-throughput DNA sequencing was performed at Massachusetts-based Life Technologies Corp.

The sequence revealed some things impossible to learn by studying the body: the color of his eyes, for example, (brown) and the fact that he was likely lactose-intolerant. But more importantly, it also gave clues to where his ancestors lived and how humans may have migrated across Europe during the Copper Age, which started about 7,000 years ago. The answer surprised some people:

“The [Iceman](#)'s ancestry most closely mirrors that of modern-day Sardinians,” said Underhill, who, with Bustamante, came to the conclusion by analyzing the mummy's Y chromosome. “His lineage is very rare in mainland Europe — only 1 percent or less share the same sequence — but is rather frequent in northern Sardinia and southern Corsica.”

Sardinia is the second-largest island in the Mediterranean Sea. It lies 120 miles west of Italy's mainland and 7.5 miles south of the French island of Corsica.



The mummy of the “Iceman” is kept in a refrigerated cell in the South Tyrol Museum of Archaeology. Credit: ©South Tyrol Museum of Archaeology

“When we found the Iceman was most closely related with modern Sardinians, it was at first hard to believe,” said Bustamante. “It’s a real mystery. Did he or his ancestors travel to the Alps from Sardinia, or did the Europe of 5,000 years ago more closely resemble Sardinia and Corsica? It’s a fascinating question in part because it addresses how rapidly people spread across Europe, and how far they rambled.”

Obtaining and sequencing DNA from such an ancient source was challenging. “Ancient DNA, which has been exposed to the elements for thousands of years, is plagued by contamination both from the environment and anyone who has handled Otzi since his [discovery](#),” said Timothy Harkins, PhD, of Life Technologies who led the sequencing effort. To limit contamination, researchers used a long needle to tap the inner part of the femur.

As it was, the researchers obtained only about 20 nanograms of genomic DNA for sequencing, which is hundreds of times less than the amount usually used for whole-genome sequencing of modern-day samples. From this, the scientists were able to identify about 2 million unique

sequence variants for population studies. One small variation on the Y chromosome pointed researchers to Otzi's island heritage.

The finding suggests two scenarios: either the mummy's ancestors were once more prevalent in mainland Europe than they are now (and some unknown selection process caused them to die off everywhere but the island strongholds), or they actively immigrated to the mainland from the island. Because there is little archeological evidence of the large, rapid population change required in the first possibility, the Stanford researchers favor the second.

"It's thought that Sardinia was first peopled about 11,000 years ago by sedentary hunter-gatherers," said Underhill. "Some samples of volcanic glass, or obsidian, found in mainland Italy and southern France have been shown to come from Mount Arci in Sardinia. This implies that there were episodes of trading between the island and mainland." If so, the mummy's ancestors could have arrived in Europe as traders.

Underhill and Bustamante are currently participating in the 1,000 Genomes Project — an international collaboration to catalog human genetic diversity. As the scientists identify increasing numbers of population-specific variants, it may become easier to track human migration and even answer questions about Otzi and other ancient specimens. "We're sequencing both modern and ancient genomes," said Bustamante, adding that technological advances have made it possible to perform analyses that would have been prohibitively expensive and time-consuming when the mummy was found some 20 years ago. "Finding these types of well-preserved specimens is by far the most labor-intensive part of the process now."

In addition to indicating [Otzi's](#) ancestry, the analysis also suggested that the man suffered from a modern malady: Lyme disease. Traces of the DNA of the bacteria that causes Lyme disease were found in the

mummy's bone marrow. It's the earliest documented case of Lyme disease in humans.

“These discoveries put a lot of what we see today medically in a broader context,” said Bustamante. “For example, this man, who died when he was about 45 years old, was likely very fit. He got lots of exercise and ate a true paleo diet. And yet he had begun to develop heart disease. This shows that the selective forces we're familiar with today just weren't in the picture then. These types of disease were probably common, but they didn't kill people. People died instead of an arrow in the back or in hunting accidents. There is still so much more to learn.”

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