



Initial genetic analysis reveals Ötzi predisposed to cardiovascular disease

Scientific magazine “Nature Communications” publishes new findings about physiognomy, ethnic origin and predisposition towards illness of the world’s oldest glacier mummy

Roughly eighteen months ago, a team of scientists succeeded in decoding the full genome of Ötzi, the mummified Iceman, revealing his entire genetic make-up. Thus the course was set for solving further mysteries surrounding the world’s oldest glacier mummy. And now the next milestone has been reached: researchers from the Institute for Mummies and the Iceman at the European Academy of Bolzano/Bozen (EURAC), and from the Institutes for Human Genetics at the University of Tübingen and Saarland University have analysed various aspects of the raw data gained from the DNA sequencing. Their findings have now been published in the scientific magazine “Nature Communications”.

Ötzi was genetically predisposed to cardiovascular diseases, according to recent studies carried out by the team of scientists working with Albert Zink and Angela Graefen from Bolzano’s EURAC Institute for Mummies and the Iceman, Carsten Pusch and Nikolaus Blin from the Institute for Human Genetics at the University of Tübingen, along with Andreas Keller and Eckart Meese from the Institute of Human Genetics at Saarland University. Not only was this genetic predisposition demonstrable in the 5,000-year-old ice mummy, there was also already a symptom in the form of arteriosclerosis, or hardening of the arteries. And yet, in his lifetime, Ötzi was not exposed to the risk factors which we consider today to be the significant triggers of cardiovascular disease. He was not overweight and no stranger to exercise. “The evidence that such a genetic predisposition already existed in Ötzi’s lifetime is of huge interest to us. It indicates that cardiovascular disease is by no means an illness chiefly associated with modern lifestyles. We are now eager to use these data to help us explore further how these diseases developed” says anthropologist Albert Zink with bioinformatics expert Andreas Keller.

Apart from this genetic predisposition, the scientists were able to identify traces of bacteria from the genus *Borrelia*, which are responsible for causing infections and are transmitted by ticks. Carsten Pusch, who led the genetic investigations in Tübingen, comments: “This is the oldest evidence for borreliosis (Lyme disease) and proof that this infection was already present 5,000 years ago.”

One further aspect which particularly interested the scientists was the Iceman’s genetic. They found that Ötzi belonged to a particular so-called Y-chromosome haplogroup which is relatively rare in present-day Europe. The findings indicate that Ötzi’s ancestors had migrated from the Middle East as agriculture and cattle-breeding became more widespread. Their genetic heritage is most common today in geographically isolated areas and islands such as on Sardinia and Corsica.

The genetic investigations also revealed a wealth of further information facts about the physical appearance of the Iceman: he had brown eyes, brown hair, and suffered from lactose intolerance which meant he could not digest milk products. This finding supports the theory that, despite the increasing spread of agriculture and dairying, lactose intolerance was still common in Ötzi’s lifetime. The ability to digest milk throughout adulthood developed steadily over the next millennia alongside the domestication of animals. The full genome sequencing was supported by the National Geographic Society (USA), by Life Technologies (USA) and Comprehensive Biomarker Center (Germany).

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