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New research reveals that Ötzi the iceman was bald and probably from a farming family. What else can DNA uncover?

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Ötzi was found in 1991 in the Alps near the Italian-Austrian border. Credit: Museo Archeologico/HO

In 1991, hikers came across a body that was partially contained in ice high up in the Alpine province of South Tyrol, Italy. Initially thought to be from a recent death, the body was later discovered to be 5,300 years old—from a time known as the Copper Age.

This amazing find would subsequently become known as Ötzi the Iceman. His body and belongings were extensively studied, prompting numerous questions: what was he doing here? Where was he from? How did he live—and die?

Researchers from the Max Planck Institute for Evolutionary Anthropology in Germany have just added another piece to this jigsaw, describing the physical appearance of Ötzi based on new DNA information. They say he probably had relatively dark skin and was balding. But how reliable are these predictions and could they be used in forensics?

Much of this depends on the quality of the samples. Ötzi died in the Otzal Alps and was frozen almost immediately, remaining in the permafrost until discovery.

The body is currently stored in low temperature conditions at the South Tyrol Museum of Archaeology. His unique preservation enabled the sequencing of Ötzi's whole genome—the complete "instruction booklet" for building a human. The chemical building blocks of DNA are called bases. These are nitrogen-containing chemical compounds called adenine, thymine, cytosine and guanine, known by the letters A, T, C and G. The human genome consists of billions of these bases arranged in different sequences—making up the genetic code.

Much of the genome's DNA sequence is common to all humans, but there are places where a change from one base to another results in changes to our physical appearance.

The Ötzi paper isn't the first study that has tried to predict a person's appearance from ancient remains. King Richard III was killed in the Battle of Bosworth in 1485. When his body was discovered in 2012, under a car park in Leicester, only his bones remained. But it was enough for a team led by Turi King at the University of Leicester to extract fragments of DNA from them.

Crime scene samples

These fragments, comprising hundreds of DNA bases, were sequenced. They were able to predict his hair and eye color and he was reliably matched to a living relative—assigning a clear identity to the remains. This means that if I ate an apple and threw the core away, I could also be identified by the DNA I left on the core.

Sequencing a genome, which comprises billions of DNA bases, enables scientists to evaluate regions of the human genome that contribute to appearance. These are known as highly variable regions.

For more than 30 years, forensic scientists have looked at specific highly variable regions in DNA to match these to crime scene samples, or to relatives of a suspect or victim. So how likely is it that DNA from such a sample could accurately paint a picture of me?

Let's take facial shape. Can forensic scientists build a kind of identikit photo from a crime scene DNA sample? Some efforts have already been taken in this regard. But our understanding of the gene variants involved in face shape are incomplete.

Many of the identikit pictures built from DNA analysis alone bear a resemblance to actual images of the individuals. But when DNA is the only evidence available to build a portrait, the prediction of facial appearance can be skewed by body composition which is significantly affected by diet and lifestyle.

However, other aspects of appearance can be predicted with high accuracy: red hair, for example. Base variations in the melanocortin receptor 1 (MC1R) gene are linked to red hair, fair skin and freckling. In rarer cases, variations in two other genes HERC2 and PIGU/ASIP are also linked to red hair.

The human genome is packaged into 23 pairs of chromosomes. On chromosome 15 there are many regions which influence eye color and skin pigmentation. Eye color can be reliably predicted, with blue eye color the most accurate. Hair color can be predicted from DNA, but darker shades of hair are more accurately predicted than blonde hair.

Aside from the complications posed by hair dye, predicting blonde hair is complicated because some individuals have very blonde hair in childhood that darkens to light brown with the onset of adulthood.

Environmental factors

Several genes contribute to produce hair pigments and a spectrum of hair color is seen in humans, ranging from light blonde to black. Commercially sold laboratory kits such as Hirisplex can simultaneously evaluate several DNA regions to predict the hair and eye color from a biological sample. However, unlike eye color, hair color prediction from DNA is only of value until midlife, when the natural processes of aging lead to graying or white hair.

These processes also lead to hair loss in some people and more than 300 gene variants have been linked to baldness. Future research should determine more clearly how these gene variants affect hair density. However, stress, diet, medication, and disease, in addition to genetics, all influence hair loss.

Individual DNA bases can become chemically modified as we age. This is known as an epigenetic change. Identical twins start life with the same DNA, but as they age, some physical differences become apparent.

Some of those differences are due to DNA bases changing as cells divide but most are due to base changes caused by lifestyle and the environment. This is an exciting area of research for understanding aging and disease. It can also be used as a forensic tool to distinguish between twins.

There is currently a lot of DNA information from people of European origin, but fewer whole genomes exist from populations elsewhere. This can influence the accuracy when scientists try to predict both appearance and ancestry.

More representative data from the rest of the world will therefore enhance studies in forensic archaeology, such as the Ötzi research. It will also have implications for forensics and assist in the identification of missing individuals.

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