How researchers discovered the genetic origin of the 'unibrow' and other hair traits

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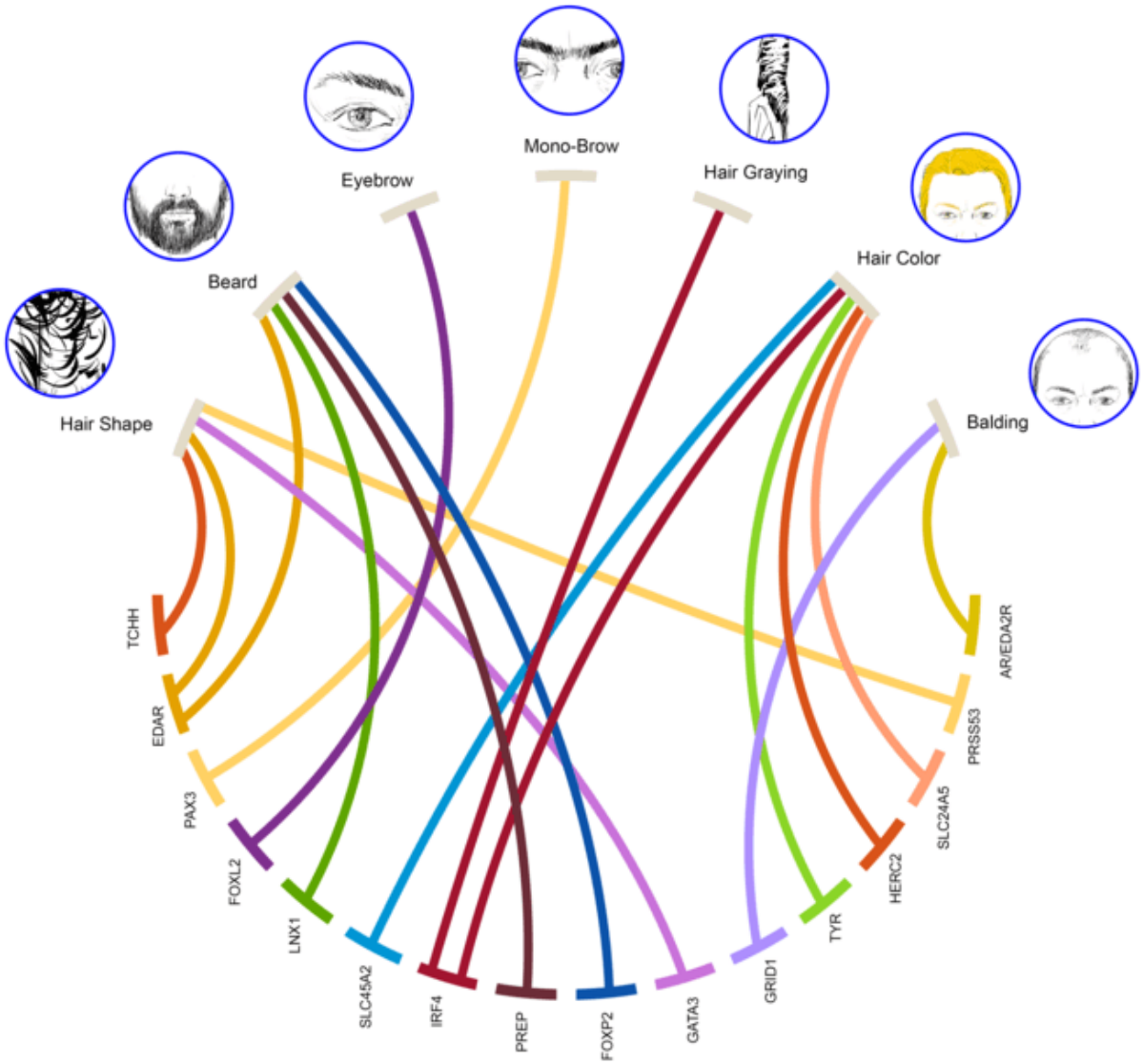
Whether we like them or not, traits such as a bald head, a "unibrow" or a distinctive hair colour are important parts of our appearance. Throughout history, people have gone through a lot of effort – and sometimes paid large sums of money – to get the style, type and colour of hair they desire. But despite our obsession with hair, scientists have long struggled to get a complete understanding of the genetic basis of hair features.

My colleagues and I have now identified a number of genetic variations associated with both scalp and facial hair – such as shape, colour and balding. While some of these traits, such as hair colour, have been linked to a few genes before, many have been completely unknown until now. The findings, published in Nature Communications, may one day help us develop drugs to alter hair growth. They also have the potential to improve forensic techniques to build pictures of suspects based on their DNA.

The Latin American melting pot

We all have preferences when it comes to hair. Some dye their hair to hide the signs of greying, while others believe that a "George Clooney look" is mature and attractive. Similarly, many people find the thick eyebrows of Mexican artist Frida Kahlo and model Cara Delevingne interesting, while others don't like them at all. Indeed, scientists have speculated that people are attracted to specific hair colours and shapes and that a process of sexual selection has led to the diversity of scalp hair patterns we see in Europe today.



Credit: Kaustubh Adhikari, Emiliano Bellini and Andres Ruiz-Linares., Author provided

However, the effect of natural selection on our hair shape is poorly understood. There are hypotheses suggesting that the predominantly tight, frizzy hair of sub-Saharan Africans evolved to provide natural insulation from the UV radiation of the tropical sun. East Asians, on the

other hand, are thought to have evolved thick, straight hair as an adaptive response to colder climates.

We often don't realise how strongly hair features are stratified across continents. For instance, straight hair is virtually non-existent in Sub-Saharan Africa, whereas we only really see variation in hair colour in West Eurasia.

We decided to carry out our study in Latin America. This is because Latin Americans have a mixed ancestry from three sources – European, Native American and African. This generates great diversity in appearance and in the underlying genes, making it easier to investigate links between them than in a more homogeneous population such as white Europeans.

For example, Europeans and Africans carry only an ancestral, unmixed variant at a particular site in a gene known as EDAR. But a mutation in this gene happened around 30,000 years ago during human migration into East Asia, which is believed to have helped these people adapt to a colder climate. This new gene variant is associated with lower sweat gland density (so heat loss due to sweating can be reduced), and thicker, straighter hair – now a characteristic of East Asians.

The results and what to do with them

Our research is based on a so called genome-wide association study of 6,630 people, designed to examine common genetic variants to see if any variant is associated with a specific hair trait. Indeed, we found EDAR to be associated with not only hair shape but also beard density. Because the derived variant in EDAR leads to lower sweat gland and hair follicle density in our body skin, it is rather expected that it would also lead to lower hair follicle in our faces – reducing hair density in beards, eyebrows and unibrows.

We identified the gene PAX3 as the unibrow gene. It has previously been shown to to control where in the face "nasion" is located – the point at the middle of two eyebrows. So it is reasonable to see it associated with unibrow, too, which is hair covering that part. Rare mutations of PAX3 have been shown to cause Waardenburg syndrome type 1 (85% of patients with this conditions have unibrow).

We also found that the gene PRSS53, which has also evolved to be different in East Asians, is involved in making hair either curly or straight, as verified through our extensive labwork. We also showed that the gene IRF4 is involved in the greying of hair by helping to regulate the production and storage of melanin, the pigment that determines hair, skin and eye colour. The figure below shows how we in this way managed to map 16 different gene variants to different hair traits.

Certainly, no gene acts in isolation, nor is one the sole determinant of any trait. These hair features are classic examples of complex traits, which may have both genetic and environmental influences – these are known as polygenic traits because multiple genes affect them.

So, now that we know all this, what can we do with it? It could be of use in forensic investigations, when trying to track somebody down from their DNA only. Current models to predict what people look like use samples from people of European descent only, but these may now finally be extended to include other populations as well.

While this is all basic research, some may have bioethical concerns about linking traits relating to appearance with genes in the long term. One such concern may be parents trying to decide the fate of a pregnancy based on their child's predicted hair colour, for example. Some have even suggested that the information could enable gene editing to give children a specific hair colour. However, we do not recommend that the results are used in this way.

The results could also have a number of cosmetic applications. By studying how the different genes involved in pigmentation work, we may be able to one day design drugs to change our hair colour "internally" without dyeing it, stop/slow balding or hair loss in general, or delay hair greying – potentially changing the way we view ourselves, and others, forever.

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