Studies of a skin color gene across global populations reveal shared origins

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All instances of a gene mutation that contributes to light skin color in Europeans came from the same chromosome of one person who most likely lived at least 10,000 years ago, according to Penn State College of Medicine researchers.

While the genetics of skin color is largely unclear, past research using zebrafish by the College of Medicine's Keith Cheng identified a key gene that contributes to lighter skin color in Europeans and differs from West Africans. In 2005, Cheng reported that one amino acid difference in the gene SLC24A5 is a key contributor to the skin color difference between Europeans and West Africans.

"The mutation in SLC24A5 changes just one building block in the protein, and contributes about a third of the visually striking differences in skin tone between peoples of African and European ancestry," said Cheng, distinguished professor of pathology. Lighter skin color may have provided an advantage due to for the better creation of vitamin D in the lesser sunlight characteristic of northern latitudes.

In this current part of the project, Victor Canfield, assistant professor of pharmacology, together with Cheng, studied DNA sequence differences across the globe. They studied segments of genetic code that have a mutation and are located closely on the same chromosome and are often inherited together. This specific mutation in SLC24A5, called A111T, is found in virtually everyone of European ancestry.

A111T is also found in populations in the Middle East and Indian subcontinent, but not in high numbers in Africans. Researchers found that all individuals from the Middle East, North Africa, East Africa and South India who carry the A111T mutation share a common "fingerprint"—traces of the ancestral genetic code—in the corresponding chromosomal region, indicating that all existing instances of this mutation originate from the same person.

These findings were reported in the journal *G3*.

The pattern of proportions of people with this lighter skin color mutation indicates that the A111T mutation occurred somewhere between the Middle East and the Indian subcontinent.

"This means that Middle Easterners and South Indians, which includes most inhabitants of India, Pakistan and Bangladesh, share significant ancestry," Cheng said.

This mutated segment of DNA was itself created from a combination of two other mutated segments commonly found in Eastern Asians—traditionally defined as Chinese, Japanese and Korean.

"The coincidence of this interesting form of evidence of shared ancestry of East Asians with Europeans, within this tiny chromosomal region, is exciting," Cheng said. "The combining of segments occurred after the ancestors of East Asians and Europeans split geographically more than 50,000 years ago; the A111T mutation occurred afterward."

Cheng plans to next look at more genetic samples to better understand what genes play the most important role in East Asian skin color. He will then use zebrafish to test those suspected genes.

The differences in skin color affect skin cancer rates. Europeans have 10

to 20 times more instances of melanoma than Africans. However, despite also having lighter skin, East Asians have the same melanoma rates as Africans. The reason for this difference can only be explained after the gene mutations for both groups are found. This understanding could lead to better treatments for melanoma.

Provided by Pennsylvania State University

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