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Different genes behind same adaptation to thin air

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Highlanders in Tibet and Ethiopia share a biological adaptation that enables them to thrive in the low oxygen of high altitudes, but the ability to pass on the trait appears to be linked to different genes in the two groups, research from a Case Western Reserve University scientist and colleagues shows.

The adaptation is the ability to maintain a relatively low (for high altitudes) level of hemoglobin, the oxygen-carrying protein in red blood cells. Members of ethnic populations - such as most Americans - who historically live at low altitudes naturally respond to the thin air by increasing hemoglobin levels. The response can help draw oxygen into the body, but increases blood viscosity and the risks for thrombosis, stroke and difficulties with pregnancies.

By revealing how populations can live in severe environments, the research may provide insight for managing high-altitude sickness and for treating low blood-oxygen conditions such as asthma, sleep apnea, and heart problems among all people.

How long such physiological and genetic changes take remains a question. The researchers found the adaptation in an ethnic group that has lived high in mountains of Ethiopia for at least 5,000 years, but not among a related group that has lived high in the mountains for 500 years.

The findings are reported today in the open-access online journal PLoS Genetics.

In their first comparison, the researchers found that the genes responsible for hemoglobin levels in Tibetans don't influence an ethnic group called the Amhara

The Amhara have lived more than a mile high in the Semien Mountains of northern Ethiopia for 5,000 to 70,000 years. A different variant on the Amhara genome, far away from the location of the Tibetan variant, is significantly associated with their low hemoglobin levels.

"All indications are we're seeing convergent evolution," said Cynthia Beall, professor of anthropology at Case Western Reserve University and one of the leaders of the study. Convergent evolution is when two separate populations change biologically in a similar way to adapt to a similar environment yet use different mechanisms.

"These were two different evolutionary experiments," Beall said of the mountain dwellers in Tibet and Ethiopia. "On one level—the biological response—they are the same. On another level—the changes in the gene pool—they are different."

Beall investigated the adaptations and genetic links with Gorka Alkorta-Aranburu, David Witonsky, Jonathan K. Pritchard and Anna Di Rienzo, of the University of Chicago department of human genetics, and Amha Gebremedhin of Adis Ababa University's department of internal medicine in Ethiopia.

In addition to studying the Amhara, the researchers looked for changes in physiology and genetics among a related ethnic group, the Oromo, who have lived more than a mile above sea level in the Bale Mountains of southern Ethiopia for 500 years.

They found no long-term adaptation and no genetic changes related to a low-oxygen environment.

They found the Omoro had high levels of hemoglobin, as would be expected for a lowland population.

Using the same samples collected from the Amhara and Oromo, the researchers are now studying biological traits among the groups, including ventilation, and the influence of vasoconstrictors and vasodilators on blood flow, and searching for associations with genes.

They also plan to continue research and study blood flow, especially through the heart and lungs of the highlanders, and to test the metabolic rate of mitochondria that use oxygen to create the energy on which our cells and we operate.

"We also want to find whether people with the variants for low hemoglobin levels have more children and a higher survival rate," Beall said. "That's the evolutionary payoff."

Provided by Case Western Reserve University