What do high-altitude Andean communities and deep-sea fish have in common? Genetic mutations that aid survival in low-oxygen environments

Living at high altitudes for long periods can be detrimental to most people's health — however, over thousands of years, some populations in the Andes and mountains of Tibet have adapted to the low-oxygen environments with genetic changes that allow them to thrive.

The same adaptation can also be seen in a deep-sea-dwelling fish.

In a new study published [February 9] in the journal <u>Science Advances</u>, researchers identified a genetic mutation in the gene EPAS1 in a group of Indigenous Quechua people in the Peruvian Andes. The mutation lowers the amount of hemoglobin — the body's key oxygen-carrying molecule — in the blood.

Mutations in this same gene have been previously tied to lower hemoglobin levels in certain <u>Tibetan highlander populations</u>. The new study highlights the importance of EPAS1 in regulating how human cells react to low oxygen levels, and it also presents a novel example of <u>convergent evolution</u> in humans, in which different populations independently evolve similar traits.

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"Thinking about how people do well in response to low oxygen, and also how people maybe don't do as well, I think is really important in a clinical context," [Tatum Simonson, an associate professor of medicine at the University of California, San Diego] said, "because we know there are people who suffer from lung disease or cardiopulmonary or cardiorespiratory diseases that respond differently to that pathological stress."

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