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Tibetan people have multiple adaptations for life at high altitudes

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Tibetan Plateau in Qinghai. Credit: DaiLuo, Flickr, CC BY

The Tibetan people have inherited variants of five different genes that help them live at high altitudes, with one gene originating in the extinct human subspecies, the Denisovans. Hao Hu and Chad Huff of the

University of Texas, Houston, and colleagues report these findings in a new study published April 27th, 2017 in *PLOS Genetics*.

The people of Tibet have survived on an extremely high and arid plateau for thousands of years, due to their amazing natural ability to withstand low levels of oxygen, extreme cold, exposure to UV light and very limited food sources. Researchers sequenced the whole genomes of 27 Tibetans and searched for advantageous genes. The analysis identified two genes already known to be involved in adaptation to high altitude, EPAS1 and EGLN1, as well as two genes related to low oxygen levels, PTGIS and KCTD12. They also picked out a variant of VDR, which plays a role in vitamin D metabolism and may help compensate for vitamin D deficiency, which commonly affects Tibetan nomads. The Tibetan variant of the EPAS1 gene originally came from the archaic Denisovan people, but the researchers found no other genes related to high altitude with Denisovan roots. Further analysis showed that Han Chinese and Tibetan subpopulations split as early as 44 to 58 thousand years ago, but that gene flow between the groups continued until approximately 9 thousand years ago.

The study represents a comprehensive analysis of the demographic history of the Tibetan population and its adaptations to the challenges of living at high altitudes. The results also provide a rich genomic resource of the Tibetan population, which will aid future genetic studies.

Tatum Simonson adds: "The comprehensive analysis of whole-genome sequence data from Tibetans provides valuable insights into the genetic factors underlying this population's unique history and adaptive physiology at high altitude. This study provides further context for analyses of other permanent high-altitude populations, who exhibit characteristics distinct from Tibetans despite similar chronic stresses, as well as lowland populations, in whom hypoxia-related challenges, such those inherent to cardiopulmonary disease or sleep apnea, elicit a wide-

range of unique physiological responses. Future research efforts will focus on identifying the interplay between various adaptive versus non-adaptive genetic pathways and environmental factors (e.g., hypoxia, diet, cold, UV) in these informative populations to reveal the biological underpinnings of individualized physiological responses."

More information: Hu H, Petousi N, Glusman G, Yu Y, Bohlender R, Tashi T, et al. (2017) Evolutionary history of Tibetans inferred from whole-genome sequencing. *PLoS Genet* 13(4): e1006675. DOI: 10.1371/journal.pgen.1006675