

Some of the ways Tibetans have evolved to stand living at such high altitudes revealed

April 4 2017, by Bob Yirka



Yarlung Tsangpo river in Tibet. In India becomes very wide and is called Brahmaputra. Credit: Wikipedia

(Phys.org)—A combined team of researchers from the University of Queensland in Australia and Wenzhou Medical University in China has identified some of the genes common to people living in Tibet that are different than people living in other areas. In their paper published in the *Proceedings of National Academy of Sciences*, the group describes the



genetic differences they found and how some of are adapted for survival at such a high altitude.

At high altitudes, there is less air for people to breath and more <u>ultraviolet radiation</u>, two factors that make living in such places rather difficult. But as prior studies have shown, people have been living in Tibet (with average elevations of 1200 meters) for thousands of years. To better understand how they survive, the researchers obtained gene samples of 3008 people living in Tibet and compared them with 7287 people that do not live at such high altitudes.

In studying the gene samples, the researchers identified nine gene differences between the Tibetans and people from elsewhere. The team found EPAS1 and ELGN1, which were identified in prior work and which were found to play a role in causing Tibetans to have lower levels of hemoglobin, which boosts the use of oxygen in the blood. But they also found ADH7, which has been associated with higher weight; this could help people survive through lean times. They also express MTHFR, which helps when nutrients are low by boosting production of folate, which is important for pregnant women. They also found a gene called HLA-DQB1, which is part of a larger group that is known to regulate proteins needed by the immune system. This would help stave off illnesses during lean times when people normally become more susceptible to diseases.

They also found four other gene differences, but since it is not clear what they do, it is not known how they might help the Tibetans survive in their <u>high-altitude</u> home. The genetic data also helped to more closely identify the time period during which the Tibetan people split from the Han Chinese population—approximately 4725 years ago, which is 2000 years earlier than other research has suggested.

More information: Jian Yang et al. Genetic signatures of high-altitude



adaptation in Tibetans, *Proceedings of the National Academy of Sciences* (2017). DOI: 10.1073/pnas.1617042114

Abstract

Indigenous Tibetan people have lived on the Tibetan Plateau for millennia. There is a long-standing question about the genetic basis of high-altitude adaptation in Tibetans. We conduct a genome-wide study of 7.3 million genotyped and imputed SNPs of 3,008 Tibetans and 7,287 non-Tibetan individuals of Eastern Asian ancestry. Using this large dataset, we detect signals of high-altitude adaptation at nine genomic loci, of which seven are unique. The alleles under natural selection at two of these loci [methylenetetrahydrofolate reductase (MTHFR) and EPAS1] are strongly associated with blood-related phenotypes, such as hemoglobin, homocysteine, and folate in Tibetans. The folate-increasing allele of rs1801133 at the MTHFR locus has an increased frequency in Tibetans more than expected under a drift model, which is probably a consequence of adaptation to high UV radiation. These findings provide important insights into understanding the genomic consequences of highaltitude adaptation in Tibetans.

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