Examining the source behind Sherpa mountain fitness

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The Sherpa population in Tibet is world-renowned for their extraordinary high-altitude fitness, as most famously demonstrated by Tenzing Norgay's ability to conquer Mount Everest alongside Sir Edmund Hillary. The genetic adaptation behind this fitness has been a topic of hot debate in human evolution, with recent full genome sequencing efforts completed to look for candidate genes necessary for low oxygen adaptation. However, few have looked at the Sherpa population by sequencing their mitochondrial genomes—-the powerhouse of every cell that helps determine the degree of respiratory fitness by providing 90 percent of the human body's energy demand, as well as controlling the metabolic rate and use of oxygen.

Unlike genomic DNA, the mitochondrial genome is unique inherited only through the mother, is small in size, and has a high mutation rate. Researchers Longli Kang, Li Jin et al. have sequenced 76 Sherpa individuals' complete mitochondrial genomes living in Zhangmu Town, Tibet, and found two mutations that were specific to the Sherpa population. The authors suggest that variants for one recent mutation in particular that was introduced into the Sherpa population about 1,500 years ago, A4e3a, that may be an important adaptation for low oxygen environments, or hypoxic conditions. This mutation is found in an "entry enzyme" stage in the mitochondrial respiratory complex, which may explain the importance of the role of mitochondria in the Sherpa population's ability to adapt to the extreme Himalayan environment.

The authors also shed light on the demographic history of Sherpa

population size over evolutionary time, showing a significant expansion from 3,000 to 23,000 around 50,000 years ago, followed by a very recent bottleneck in the past several hundred years that reduced the population from 10,000 to 2,400, matching known historical migration patterns.

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