

## Evidence found of gene markers that may help people survive at high altitudes

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A team of researchers with members from several institutions in the U.S., one from Peru and one from India, has found evidence of gene markers in native Andean people that may help them survive at high



altitudes, where oxygen levels can sometimes be half as much as lowland areas. In their paper published in *Proceedings of the National Academy of Sciences*, the group describes their study, which involved comparing part of the genome of highland people with lowland people.

Prior studies have shown that the reason people who live high in the Andes mountains for generations have enhanced exercise capacity is partly because they have larger-than-average lungs. They also typically have higher aerobic capacity and more hemoglobin in their blood. And now, it turns out they may have a genetic advantage, as well.

To better understand why natives of high mountain communities are able to live and even carry out physically taxing activities, the researchers carried out a genetic study of 429 people who live in the highlands of Peru and compared them with the genes of 94 people living in a lowland part of New York. The team looked specifically at the EGLN1 gene in the volunteers because it has been associated with possible mutations that might help people live at high altitude. The researchers report that they found five genetic markers that were different between the two groups of volunteers. They also note that the variants have been associated in the past with conferring a higher aerobic capacity in hypoxia, a finding that suggests those with the marker are better able to handle low oxygen levels in the air. They further note that the EGLN1 gene is known to play a role in the hypoxia-inducible factor complex, in which the gene encodes for a protein that controls the means by which cells respond to low oxygen levels.

The researchers acknowledge that their study did not pinpoint a gene involved with heightened athletic performance, but their findings did suggest that genetics may play a role in helping some people to better deal with low-oxygen environments.

More information: Tom D. Brutsaert et al. Association of EGLN1



gene with high aerobic capacity of Peruvian Quechua at high altitude, *Proceedings of the National Academy of Sciences* (2019). <u>DOI:</u> <u>10.1073/pnas.1906171116</u>

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